PRENATAL GENETIC SCREENING FOR CYSTIC FIBROSIS CARRIERS: IMPLICATIONS FOR MATERNITY CARE

Moira E Mennie

Thesis submitted to

FACULTY OF SOCIAL SCIENCES

UNIVERSITY OF EDINBURGH

for the degree of

DOCTOR OF PHILOSOPHY

December 1994

DECLARATION

Excerpts from the results presented in this thesis have been published as detailed on page xiv. I certify that this thesis does not contain any other material published or written by any other person except where due reference is made in the text. The results presented in this thesis have not been submitted for any other degree or diploma.

MOIRA E MENNIE

APPENDIX

1. The information leaflet issued to all women with their booking clinic appointment, entitled: Cystic Fibrosis Carrier Testing Prenatal Screening Trial.

2. The pre-screening questionnaire including the 12-item General Health Questionnaire.

3. The Symptom Rating Test (SRT)

4. The Facts and Feelings Questionnaire

5. Published papers

ACKNOWLEDGEMENTS

I would like to thank the following people:

Professor DJH Brock for providing me with the opportunity to undertake this project, and for his encouragement throughout the course of this study.

Dr DA Whyte and Dr I Pullen for supervising this project and for their guidance and encouragement, thoughtful suggestions and patience in reviewing early drafts of this thesis.

My colleagues Sister Mary Compton and Miss Annette Gilfillan who gave me the kind of professional and personal support for which no expression of thanks could be adequate.

Dr WA Liston for accommodating me throughout the four years it has taken to complete this thesis and for his humorous, down-to-earth and wise comments.

The antenatal clinic staff of the Simpson Memorial Maternity Pavilion without whom this work would not have been possible.

The many women and their partners whose thoughts, opinions and feelings embody this thesis.

The Cystic Fibrosis Trust for financing this research.

PUBLICATIONS

Excerpts from the results presented in this thesis have been published as detailed below.

Mennie ME, Liston WA, Brock DJH. (1992) Prenatal cystic fibrosis carrier testing: designing an information leaflet to meet the specific needs of the target population. J Med Genet 29: 308-312.

Mennie ME, Gilfillan A, Compton M, Curtis L, Liston WA, Pullen I, Whyte DA, Brock DJH. (1992) Prenatal screening for cystic fibrosis. Lancet 340: 214-216.

Mennie ME, Compton ME, Gilfillan A, Liston WA, Pullen I, Whyte DA, Brock DJH. (1993) Prenatal screening for cystic fibrosis: psychological effects on carriers and their partners. J Med Genet 30: 543-546.

Mennie ME, Gilfillan A, Compton ME, Liston WA, Brock DJH. (1993) Prenatal cystic fibrosis carrier screening: factors in a woman's decision to decline testing. Prenat Diagn 13:807-814

Mennie M, Compton M, Gilfillan A, Axton RA, Liston WA, Pullen I, Whyte DA, Brock DJH. (1993) Prenatal screening for cystic fibrosis: attitudes and responses of participants. Clin Genet 41:102-106

Mennie M.E. (1994) Prenatal screening for cystic fibrosis. Br J Midwifery (in press)

ABSTRACT

The aim of this research was to measure and describe the impact of screening for cystic fibrosis (CF) carriers in pregnancy and to assess the implications for midwifery practice. Women were invited to be screened by an information leaflet sent with their booking clinic appointment. The midwife booking a woman at the clinic offered further information and counselling. Male partners of women identified as carriers were also invited to be screened and heterozygous couples were offered prenatal diagnosis.

A cohort of 2,058 women offered screening completed a self administered questionnaire which measured their knowledge and attitude to screening and described their reasons for accepting or declining the test. Psychological status, prior to testing, was measured by the General Health Questionnaire (GHQ) and the Symptom Rating Test (SRT).

Women 16-20 years were significantly less likely to have heard of CF; significantly more likely to find the information leaflet difficult; and significantly less likely to have a partner who read the leaflet or to discuss the test with him. Women 16-25 years were significantly less likely to know their risk of being a CF carrier. 27% of all women discussed the test with no one other than the midwife. 25% felt anxious about being screened. 62% of those accepting, wished to determine fetal CF status, of whom 10% wished to prepare for an affected child. 54% who declined were opposed to abortion - (43% totally and 11% specifically for CF). 30% of those totally opposed accepted maternal serum alpha-fetoprotein (MSAFP) screening. 32% of women showed signs of psychological disturbance prior to screening.

The psychological response of 64 women identified as CF carriers and their partners who received a negative test result were assessed together with selected controls on 4

further occasions: 1) on receiving the carrier's positive test result; 2) on receiving the partner's negative test result; 3) six weeks later; 4) six weeks after delivery. Knowledge of the genetics of CF and attitude to having been screened were measured by self administered questionnaire. Compared to control subjects carriers showed a significant increase in generalised psychological disturbance attributed specifically to symptoms of anxiety and depression during the period awaiting their partner's test result but returned to control levels on receipt of a partner's negative test result. Although there was no significant difference in generalised psychological disturbance between partners and their selected controls, partners did become significantly more anxious and manifested signs of inadequacy while awaiting their own test result.

All four groups were well informed about the genetics of CF and the significance of being a gene carrier, although 23% of carriers felt information given at the booking clinic was insufficient. 20% of carriers felt regret or ambivalence about having been screened. There was a consensus that screening should be routinely offered to pregnant women but should also be made available in family planning clinics and GP centres.

Results showed that the implications for midwifery practice focus on 3 areas of care: information giving; counselling; and emotional support. Practical guidelines for the presentation of screening within these three areas are outlined.

Concerns which arise are: current irregularities in the presentation of screening tests; genetic screening focusing on pregnancy and placing women in an unfair position of responsibility for genetic disease; information overload causing blockage or misinterpretation of other antenatal health care messages; the expansion of genetic screening tests could cause an imbalance of antenatal information toward the abnormal rather than the normal.

Notes

The use of the female pronoun when referring to midwives was not intended to exclude those midwives who are male.

All names of women or their partners used in the text are fictional.

Permission to use the General Health Questionnaire (GHQ) and The Symptom Rating Test (SRT) from NFER-NELSON.

Permission to include photocopies of publications from:
Journal of Medical Genetics, BMA House, Tavistock Sqare, London.
Clinical Genetics, Munksgaard International Publishers Limited, Copenhagan.

CHAPTER 1
INTRODUCTION

INTRODUCTION

When a child is born with a genetic disease parents search for the presence of a family history. For over 50 years cystic fibrosis (CF) has been recognised as a genetic disease. In Britain, about one child is born each day with CF; yet, in 4 out of 5 affected infants the parents have no family history of the disease (Boat et al 1989). Understandably, these parents ask "why us?"

Parents of a child with CF are, by definition, obligate CF carriers. They have no symptoms of the disease but, with each pregnancy they have a 1 in 4 chance of having a child with CF. Until recently there was no way to detect the presence of the CF gene in a family before an affected child was born. Then, in 1989 the gene which causes CF was identified on chromosome number 7 (Rommens et al 1989; Riordan et al 1989). Following this discovery the development of a test to detect carriers of the CF gene meant that individuals in the general population could be screened and couples at risk of having a child with CF could be identified. Since 1 in 25 individuals in the United Kingdom carry a CF gene (Boat et al 1989), the test had the potential to detect a majority of the 1 in 625 couples at risk of having an affected child. These couples could then be offered prenatal diagnosis to detect the presence or absence of the disease in their unborn child.

Immediately important questions arose concerning population carrier screening for CF. Could and should population carrier screening be implemented? If so should it be offered selectively or universally, and at what stage - birth, school, pre-pregnancy or prenatally? What would the level of uptake and public attitude be to such screening? Although genetic screening programmes existed for the genetic disorders beta-thalassaemia, sickle cell disease and Tay Sachs disease they were restricted to the ethnic

minority groups in which these disorders are prevalent. Thus, there was no precedent for a large scale genetic screening programme in the United Kingdom. The consensus was that pilot programmes were urgently needed to investigate these fundamental questions about CF carrier screening. Consequently, the Cystic Fibrosis Trust invited interested parties to submit protocols for screening trials for their approval and financial support. Three pilot projects were selected for funding by The Trust, each designed to examine differing approaches to carrier screening. In Cardiff and London the feasibility of screening in general practice was assessed. In Edinburgh, the director of the third programme advocated a prenatal approach to screening based on the belief that pregnant couples coming through an antenatal clinic are more motivated and more in need of this type of information and more likely to make immediate use of the information provided through screening (Brock et al 1991). In addition, the concept of maternal serum alpha-fetoprotein screening delivered by midwives had been successfully initiated by the director in the same hospital as that in which the CF carrier screening trial was to operate (Brock and Sutcliffe 1972). Thus, there were already in place midwives experienced in the presentation of prenatal screening.

The researcher was appointed as co-ordinator of the prenatal CF carrier screening trial. Her role was to design and evaluate a screening protocol and clarify what information should be provided to the target population and evaluate ways of presenting that information. In addition, she was required to design a study which would answer the question: 'is this form of prenatal screening acceptable to women and their partners?' It is this study which forms the broad substance of this thesis.

Since the prenatal CF carrier screening trial was to be presented by midwives it seemed appropriate to approach the study from this perspective. Nursing and midwifery has a commitment to the philosophy of holistic care. Nurses and midwives recognise that a

person can be viewed as having psychological, social, spiritual and physical domains and that when problems arise in any one of these domains it affects the individual and significant others in one or more of the other domains (Price 1990). Effective care depends upon nurses and midwives having the knowledge and the ability to assess and diagnose before they can plan, implement and evaluate interventions to help the individual. Presently it is not possible to suggest ways that midwives might best help those who undergo genetic screening because we have little or no understanding of what, if any, the range of associated problems might be. Before the profession can consider taking responsibility for the delivery of prenatal genetic screening it needs to acquire information relevant to a midwifery perspective of the screening process.

Research has been defined as: "an attempt to increase the sum of what is known, usually referred to as 'a body of knowledge' by the discovery of facts or relationships through a process of systematic scientific enquiry, the research process." (Hockey 1991 page 4). Nursing research aims to increase the sum of what is known about the professional activity of nurses, which may be nurse education, nursing administration or nursing practice in its many forms and settings (Hockey 1991). The purpose of research is to develop theories for nursing and midwifery practice which establishes the association between individual or patient needs, nursing intervention and individual or patient outcome (McFarlane of Llandaff 1991).

This thesis considers the consequences to women and midwives of offering genetic screening in the context of an antenatal clinic. It describes and examines factors which emerged from studying a cohort of approximately 2,500 women who were offered prenatal CF carrier screening and 64 women who were identified as CF carriers.

The broad aim of the work was to contribute to the body of knowledge about the midwife's role in presenting prenatal genetic screening.

The thesis begins (Chapter 2) by outlining the background to the study. It describes the disease cystic fibrosis (CF) and the development of a test to screen for CF carriers in the general population. It reviews genetic screening tests and pregnancy screening tests, and describes a trial of a prenatal model of CF carrier screening which presented the opportunity to undertake this research. From this description of the background to the study a number of key questions emerge relating to midwifery care and these are addressed. A major concern is that screening may cause distress which could persist throughout pregnancy.

Chapter 3 focuses on the subject of stress, in response to initial concerns that the major impact of prenatal CF carrier screening was likely to be a psychological one. Models of stress and coping are reviewed and the conceptual framework for the study is drawn from a model of stress, coping and mental health. The model is used to formulate an outline for a more extensive literature review.

Chapter 4 is devoted to the literature review which focuses on a number of key areas which were identified using the conceptual model. These key areas of genetic screening, genetic disease, prenatal screening, early infant loss, pregnancy and concurrent life events are reviewed in relation to their impact on an individual or couple. The conclusions drawn are used to frame a number of broad based questions relating to prenatal genetic screening .

Chapter 5 deals with the process of refining these broad questions in the context of the conceptual model. The research methodology selected to conduct the study is outlined.

The questions asked are:

1 Are there pre-screening variables which influence a woman's response to prenatal CF screening?

2 What factors influence a woman to accept or decline prenatal CF carrier screening?

3 Will identifying a woman as a CF carrier during pregnancy provoke a stressful response both in her and her partner?

4 Do carriers and their partners understand the essential facts concerning CF carrier screening and what is their attitude toward having been screened?

Chapter 6 presents the results of the study by addressing each question in turn and discussing the findings.

Chapter 7 focuses on how the results of the study contribute to nursing knowledge about the midwife's role in prenatal genetic screening. Firstly a number of ethical issues which emerged from the study are discussed; the intent being to create an awareness of these ethical themes which thread through the whole prenatal genetic screening process. The conceptual model is used to provide a framework to assess how the results of the research contribute to midwifery practice. Practical guidelines for the presentation of prenatal genetic screening are proposed on the basis of the results. Implications for midwifery managers and educationalists are discussed and suggestions are made for further research. Finally, a number of concerns about using a prenatal model of genetic screening are addressed.

CHAPTER 2

BACKGROUND TO THE STUDY

BACKGROUND TO THE STUDY

2.1 Cystic fibrosis

Cystic fibrosis (CF) is a multi-system disease, first described in the 1930s (Andersen 1938). It affects the respiratory, gastrointestinal and reproductive systems, as well as the sweat glands. It is the most common life shortening, recessive genetic disorder among Caucasians of European descent, occurring in 1 in 2,500 live births in the United Kingdom (Beaudet 1989). The disease does occur in other races but at a lower incidence. Cystic fibrosis varies in severity from patient to patient and to the extent to which different organs are affected. There is no cure for CF, but treatment of the respiratory and digestive symptoms has lengthened life span considerably.

2.1.1 Pathology

Although the disorder is present at birth, only 10 per cent of infants are born with a detectable symptom of the disease called meconium ileus. The baby fails to pass meconium and there is increasing abdominal distension and bileus vomiting. A few cases can be treated with gastrografin (sodium and methyl glucamine diatrizoates) enemas but the majority require surgery (Hodson 1993). By the age of 3 years most affected children have developed symptoms, although in a minority these are not apparent until later in childhood, adolescence or even adulthood (Boat et al 1989).

CF occurs because of a dysfunction of the exocrine glands. These glands secrete into ducts or onto specific organ surfaces and include the lacrimal glands, sweat glands, part of the pancreas and the mucus producing cells lining the respiratory and gastro-intestinal tract. In CF secretions from the serous glands have an increased salt content. In contrast, the mucus secreting glands have a normal or diminished salt content and, in addition,

thicker than normal mucus secretions. The latter leads to obstruction of the gland ducts resulting in the varied clinical features of the disease (Goodchild and Dodge 1985).

2.1.2 Clinical features

Respiratory system

The thick, sticky mucus produced in the respiratory tract of affected individuals obstructs breathing and interferes with the normal exchange of gases and removal of bacteria from the airways. Chronic lung infection, subsequent inflammation and resulting lung tissue damage limits pulmonary function and finally causes respiratory and heart failure. It is the severity of respiratory involvement which is the life threatening component of CF, ultimately determining quality of life and survival (Penkeith et al 1987).

Gastrointestinal system

In childhood, digestive difficulties may predominate over respiratory symptoms. Eighty five to 90 per cent of cases manifest some pancreatic involvement related to inadequate quantities of pancreatic enzymes being released to digest food (Taylor 1993). Poor nutrition and impaired growth is the result of fat and protein not being broken down and absorbed by the body. Nutritional status is now regarded as central to prognosis. Poor nutrition intensifies respiratory disease and a balanced diet with extra calories and protein, sufficient to promote normal growth and weight gain, will help the individual withstand chest infections (Taylor 1993). Diabetes occurs in approximately 12 per cent of adult CF patients. Older patients are also susceptible to bowel obstruction which can sometimes be treated orally or failing this by enema. In the majority of adult patients there is also some degree of liver involvement,

characteristically biliary cirrhosis, resulting from blockage of the ducts that transport bile into the intestine (Hodson 1993).

The reproductive system.

Cystic fibrosis manifests itself in the reproductive system of males and females. In 95 per cent of males the Wolffian duct is damaged (Boat et al 1989). The vas deferens are often absent, incompletely formed or blocked by mucus (Rigot et al 1991) and in addition it is thought that sperm may be formed imperfectly in men with CF (Trezise and Buchwald 1991). Consequently, only 2 or 3 per cent of males with CF are fertile (Boat 1989). Women with CF may produce thick dehydrated mucus which can impede sperm migration and plug the opening to the uterus thus reducing the chance of a pregnancy (Boat et al 1989). Women may also develop amenorrhoea secondary to poor nutritional status or pulmonary disease. Lastly, pregnancy imposes an added burden on the respiratory system and can result in deterioration of health in a women with CF (Cohen et al 1980). Those with poor lung function are advised to avoid pregnancy (Hodson 1993). In recent years a number of CF adults have successfully had children (Duncan-Skingle and Foster 1991). The risk of a parent with CF having an affected child is 1 in 50 (Brock 1993).

2.1.3 Diagnosis

The sweat test was until recently the most common method of confirming a suspected diagnosis of CF. Excess sodium and chloride are lost in the sweat. A raised sweat chloride confirms the diagnosis of CF along with typical clinical findings (Gibson and Cooke 1959). Sweat testing in the new-born is ineffective and the immunoreactive trypsin test (IRT), which measures levels of pancreatic trypsin, elevated in CF, is performed by Guthrie spot (Crossley et al 1979) but is not routinely offered. Direct gene analysis, now the most precise diagnostic test, is described in section 2.2.4 page 17.

2.1.4 Medical management

The medical management of CF focuses on impeding loss of lung function by daily physiotherapy to promote mucus clearance, together with appropriate bronchodilator therapy. Antibiotic therapy varies from intermittent use to repeated sustained use, however, bacterial resistance to treatment is a major problem. As the affected individual's condition deteriorates they develop chronic hypoxia and respiratory failure. At this stage heart-lung transplantation is the only option (Duncan-Skingle and Foster 1991). Both heart-lung, and double-lung replacement surgery have been performed (Wrightson et al 1993). As with all organ transplantation, rejection is the greatest problem to survival.

Digestive therapy strives to achieve ideal weight, normal growth, sustain respiratory muscle strength and maintain immunity. An important adjunct is a high calorie, high protein diet and pancreatic enzyme replacement to counteract malabsorption. Food supplements and occasionally intravenous nutrition may become necessary. Enteric coated enzymes are taken with each meal and snack, and supplementation of the diet with fat soluble vitamins is required. The nutritional needs of the CF child are fully described by Taylor (1993).

2.1.5 Prognosis

Since the arrival of antibiotics in the 1940's, the introduction of chest physiotherapy in the 1950's and advances in nutritional management in the 1970's and 1980's, the life expectancy of those with CF has increased. Some believe that earlier diagnosis and hence treatment improves prognosis, but many believe that the advent of CF centres has had the greatest single impact; demonstrated by the differences in survival between countries (Boat et al 1989; George 1990; Dodge et al 1993).

It is estimated that there are some 6,000 CF sufferers in the United Kingdom, among whom 34 per cent are aged 15 years and over. By the turn of the century the total number of affected individuals is expected to rise to around 7,000. The average life expectancy is currently estimated to be 25 years for males and 24 years for females (Dodge et al 1993). Cystic fibrosis can no longer be considered solely a paediatric problem and recommendations for the care of adults with CF have been made (Royal College of Physicians 1990). Now the identification of the CF gene promises to have an even greater impact on the treatment of this disease.

2.1.6 Burden of the disease

As with any chronic disease CF has a profound effect not only on the lives of the individual sufferers but also on their families, resulting from physical symptoms, psychological and social consequences. For the affected person a chronic productive cough, offensive stools with flatulence, short stature, delayed puberty and infertility can combine to create a life long struggle for many sufferers, resulting in depression, anxiety and feelings of stigmatisation (Smith et al 1983). Children may resist the stringency of daily physiotherapy and rebel against the constant ingestion of digestive enzymes placing a strain on the patience of parents and disrupting family life. One study reported those with CF to be more socially isolated and dependent upon their mothers than their peers (Cappelli et al 1989). This, it was claimed, was a consequence of the great amount of time and management their condition demanded. CF requires daily treatments; even mildly affected individuals require pancreatic enzymes and usually chest physiotherapy which affects the daily lives of both patient and family. Fifty per cent of mothers in one study complained about the time they spent on their affected child, leaving little time to look after the rest of their family. One-third of these mothers stated that they felt genuinely stretched to cope (Andesson-Segesten and Plos 1988). These findings are significant in view of recent work which shows that

balanced family coping and resulting compliance with therapy optimises child health outcome (Patterson et al 1993). Balanced family coping was defined by Patterson and colleagues as parents who: "tried to maintain family integration and worked together to meet the medical needs; they attended to their own personal esteem and support needs; and they sought medical consultation from doctors and others" (p 388).

Adverse emotional responses of parents to the birth of a child with CF can have a pervasive effect on the family making them vulnerable to crisis. Whyte (1992) revealed how parents, when their child is diagnosed as having CF, often respond with fear and denial. The source of these emotions came from a sense of threat which parents felt from the loss of their child's health.

As the life expectancy for sufferers of CF has improved, it has become increasingly a disease of young adulthood rather than a disease of childhood. Because CF is a progressive disease, young adults find they are increasingly less fit and yet the demands of daily therapy are just as important, if not more so than before. At a time when the emphasis is on education, employment and independence extra demands are placed upon the adolescent and young adult sufferer (Lewiston 1990; Shepherd et al 1990). Nonetheless, a recent survey reports many young adults are as healthy psychologically as their peers and are living full and productive lives (Walters et al 1993). Walters and colleagues found that 55 per cent of sufferers in their study, over the age of 16 years, were working and 56 per cent of them had less than two weeks sick leave a year. Of those not employed, 50 per cent gave ill health as the reason. Ill health caused more CF sufferers to leave school without qualifications than in the general population and a higher proportion remained within the parental home.

The average annual cost of treating someone with CF is estimated to be £4-6,000 (Office of Health Economics 1986). However, there is the inestimable non-medical cost of family care giving time. It is estimated that parents need to spend about 2 hours every day on therapy for a child with CF (United States Congress, Office of Technology Assessment, 1992a). In addition, family members lose time from work or home commitments when the CF sufferer is sick.

Death among their peer group is a reminder to CF sufferers of their own shortened life expectancy. Those who receive donor organs for lung transplantation must face another set of emotional problems such as altered body image from scarring and steroid side effects, uncertain prognosis, guilt and grief relating to the donor, as well as financial, family and employment difficulties (Wrightson et al 1993). It is understandable that the identification of the CF gene has lead to high expectations among patients, their families, physicians and nurses for dramatic improvements in treatment.

2.2 The Genetics of CF

2.2.1 The inheritance of CF

An individual has two copies of chromosome 7, one inherited from their father and one from their mother. Individuals with CF inherit a defective gene on each of their chromosomes number 7. This happens in 1 in 2,500 babies in the United Kingdom (Boat et al 1989). An individual may have a CF gene on one of their number 7 chromosomes and a normal gene on their other number 7 chromosome; these individuals are CF gene carriers but have no symptoms of the disease. One in 25 of the UK population are carriers and in 1 in 625 couples both partners are carriers. Couples where both partners are CF gene carriers are at a 1 in 4 risk at each pregnancy of bearing a child with the disease and a 2 in 3 chance that an unaffected child will be an asymptomatic carrier like themselves (Boat et al 1989) (Figure 2.1).

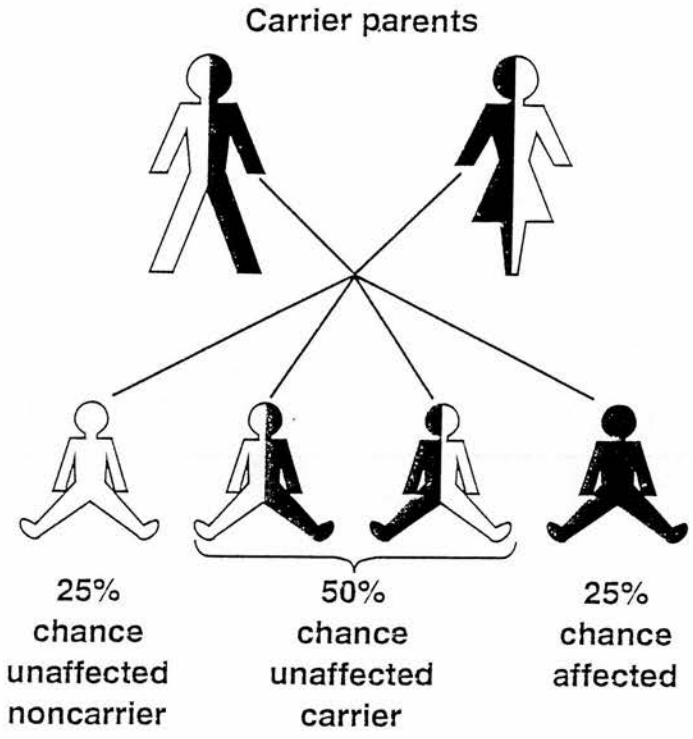


Figure 2.1 Inheritance of CF

Most couples with an affected child have no family history of the disorder and have no idea that the genetic trait exists in the family. The risk of couple having a child with CF given a variety of situations is shown in table 2.1.

Table 2.1 Risks to couples of having a CF child (adapted from Brock 1993)

Couple	Risk of CF child
Couple have one or more affected children	1 in 4
Both partners have affected siblings	1 in 9
Both partners have carrier siblings	1 in 16
One partner has an affected sibling	1 in 150
One partner has a carrier sibling	1 in 200

2.2.2 Identifying the CF gene

In genetics the search for how specific traits are passed from one generation to the next has been greatly assisted by the new technology of molecular biology. Geneticists now have techniques which will allow them to locate and isolate a single gene from one of our 46 chromosomes. Genetic diseases arise as a result of changes (mutations) in the deoxyribonucleic acid (DNA) that comprises a gene. Approximately 4,000 known human disorders result from genetic causes (McKusick 1990). Disorders which arise from a mutation in one gene are called monogenic or single gene disorders. CF is such a disorder.

Genes produce a set of instructions or molecular code which specify the manufacture of a particular protein to function in the cells of different organs. When a gene is defective or faulty it can affect the structure, regulation, function or synthesis of a protein which in turn can lead to malfunction of a particular organ. In CF the exocrine glands are affected. In 1989 the gene responsible for CF was isolated on chromosome 7 and the most common mutation identified. The product of the CF gene is a membrane-transport protein which has been named the cystic fibrosis transmembrane conductance regulator (CFTR) The tissue expression of the gene is compatible with the pathophysiology of the disease. (Riordan et al 1989).

2.2.3 Current genetic research

As the workings of the CF gene are better understood, new possibilities for treatment are emerging and gene therapy has moved from theory to clinical and therapeutic experimental application (Anderson 1992; Davies and Williamson 1993). The development of a cystic fibrosis mouse allowed copies of the normal human CFTR gene to be introduced into the lungs using liposomes (lipid vesicles which fuse with the epithelial cells), leading to the ion transport defect being corrected in some cases

(Hyde et al 1993). In 1992 the 'Clothier Committee' reported on the ethics of gene therapy and stated they found "no new ethical issues" raised by somatic gene therapy (see footnote 1). Protocols for gene therapy trials are currently being submitted for approval and funding (Davies and Williamson 1993).

2.2.4 The CF carrier screening test

The discovery of the CF gene means that scientists have been able to improve CF diagnosis, including prenatal diagnosis and devise screening tests to identify people who carry a defective copy of the gene and run the risk of having a child with the disease.

A laboratory technique, the polymerase chain reaction (PCR), has proved critical for carrier detection and diagnosis by direct gene analysis (Saiki et al 1988) (see footnote 2). Over 400 mutations have been identified in the CF gene, but many have been detected in only 1 family. Some five mutations were commonly found in the Scottish population allowing some 85 per cent of carriers to be identified. This rate of detection was considered adequate to allow a screening trial to commence (Shrimpton et al 1991).

---

1. There are two approaches to gene therapy: somatic gene therapy aims to correct the cells of specified affected organs such as the lungs in CF. There is no attempt to correct the defect in egg or sperm cells and therefore the correction is not transmitted to any children of an affected individual. In contrast, germ line gene therapy would lead to the correction of all cells in an individual including the germ line. This correction would be passed on to following generations (Coutelle et al 1993).

The remit of the Clothier Committee was to consider the ethics of gene therapy; to consider proposals for treatment; and to provide advice on safety and efficacy (The Clothier Report 1992).

2. The CF carrier test involves directly analysing the gene. DNA can be obtained from any nucleated cell in the body. White blood cells or buccal cells are most commonly used for carrier screening or diagnosis and amniotic fluid cells or chorionic villi for prenatal diagnosis. After the DNA is extracted, key segments containing mutations are amplified using the polymerase chain reaction technique (PCR). This technique amplifies specific areas of DNA to increase the amount available for test purposes. The amplified DNA can be visualised by a number of different laboratory techniques to detect a specific mutation (Saiki et al 1988).

2.2.5 Options available to carrier couples

In the case of carrier couples with a 1 in 4 risk of an affected child, prenatal diagnosis can predict with 100 per cent accuracy if the fetus is affected. The choice of technique to obtain a sample of DNA from the fetus depends upon the gestation of pregnancy and consideration of procedural risk. Fetal cells can be obtained via chorionic villus sampling (CVS) performed from 8-10 weeks gestation of pregnancy onwards. The biopsy is taken under ultrasound guidance via a transcervical route or via a transabdominal route. CVS has a procedural associated risk of spontaneous abortion of 2 to 4 per cent (Lilford 1991). An alternative method of obtaining fetal cells is by amniocentesis, the withdrawal of amniotic fluid, performed from 14 weeks gestation of pregnancy. A needle is introduced under ultrasound guidance into the amniotic cavity via the maternal abdomen. The risk of spontaneous abortion occurring as a result of amniocentesis is 1 per cent (Lilford 1991).

Some couples may decide against prenatal diagnosis and opt to continue the pregnancy without confirming the status of the fetus. It is important in those pregnancies which may result in an infant with CF to alert neonatal paediatricians to the risk of meconium ileus. Although the knowledge gained through prenatal diagnosis has the potential to offer relief from the apprehension of having an affected child, it can present agonizing dilemma among families who already have a child with CF and who by considering termination of pregnancy feel they might be seen to devalue their affected child (Whyte 1992).

Couples who are screened prior to conceiving confront a wider variety of options. Figure 2.2 outlines the differing options which pre-pregnancy and prenatal screening offer.

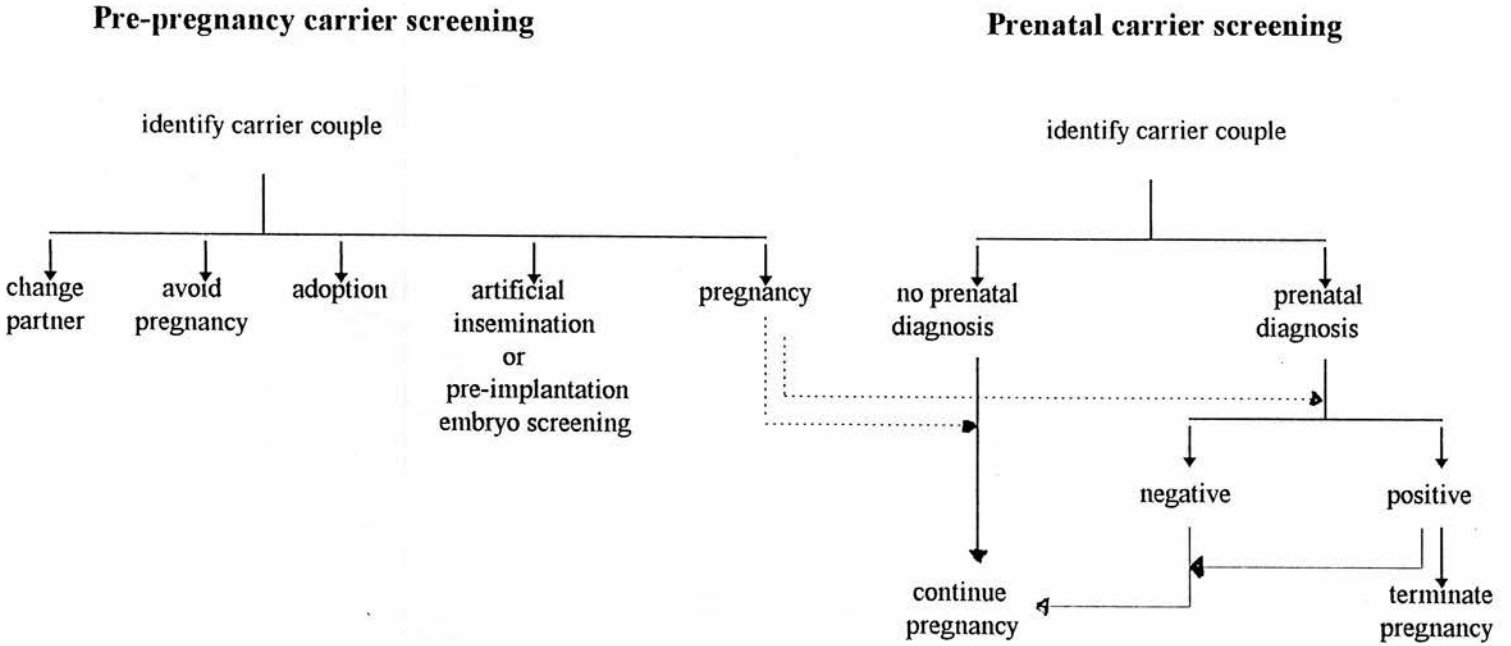


Figure 2.2 Pre-pregnancy and prenatal carrier screening: the differing options

2.2.6 The initiation of population carrier screening trials.

New technologies bring new challenges to society. Almost immediately the CF gene was identified, it became apparent that there was a realistic means of detecting many of those couples who were at risk of having a child with the most common of lethal autosomal recessive disorders, before an affected child was born. Such a development has prompted the comment that within the next 10 years genetic screening will affect the majority of people in developed countries (Wertz and Fletcher 1990). However well intentioned, implementing genetic screening programmes prematurely could cause harm and has led to recommendations that well designed pilot trials should be undertaken to address pertinent issues such as autonomy and informed consent; confidentiality, psychological effects and evaluation of outcome (Baird 1990). Before looking to the future the different types of genetic screening tests already available and those groups of individuals at whom they were targeted was reviewed.

2. 3 SCREENING

Definition of screening

Screening has become a popular concept in health care and now has wide acceptance in our society (Holland and Stewart 1990). Screening is defined as the identification among apparently healthy people of those at sufficient risk of a specific disorder to warrant tests and treatment (Cuckle 1990). A screening test is not usually a diagnostic test, for example if a cervical smear test or mammogram is positive, further investigations such as a colposcopy or biopsy are required to confirm and determine the nature of the abnormal findings. Once the diagnosis is confirmed treatment or preventive advice or action can be offered (Chapple 1992).

2.3.1 Genetic Screening

Screening for the disease state

Some genetic screening tests determine whether an individual carries a gene which will cause a specific disease. Screening programmes for a number of genetic diseases have been running for many years using tests which detect a metabolic product, for example, neonatal screening for phenylketonuria in which a raised phenylalanine level is detected in the blood of infants. The benefit is to the individual who can then be treated and mental retardation avoided (Guthrie 1968). More recently, as genes that cause certain diseases have been identified, it is possible to diagnose the disease state by the presence of the mutant genes, for example, cystic fibrosis in a child suspected of having the disorder (Rommens 1989). A further use of genetic testing is in screening individuals at high risk of developing a genetic disorder because of a family history, to determine if they are negative or positive for the disease state before they manifest symptoms of the disorder. Presymptomatic genetic testing for late onset genetic disorders such as Huntington's disease and myotonic dystrophy requires very careful pre-test counselling and follow-up support (Harper 1984).

Genetic carrier screening

In contrast, genetic carrier screening does not test for the disease state; rather it identifies individuals who carry one normal and one aberrant copy of a gene, but not the disorder which results from having two aberrant copies of the gene. Cystic fibrosis is an example of such a recessive disorder where carriers can be identified either from the general population or within families where there is a family history. There is no direct benefit to the individual from carrier screening, rather it allows them to make reproductive choices (Chapple 1992). These choices have been outlined in figure 2.2.

There are a number of turnstiles in life when screening can easily be offered. Birth is an efficient time to carry out screening and many countries screen for phenylketonuria using blood spots which test almost 100 per cent of infants, and treatment for the disease can then be initiated (Guthrie 1968). However, screening for CF carriers at birth provides information which is not relevant to the child until reproduction.

Alternatively, screening in high schools could be easily administered and combined with an educational programme. Pilot projects in Canada indicate minimal problems, providing there is careful counselling and support available (Kaplan 1992).

During pregnancy, screening can be offered to a woman and if she tests positive her partner can be screened. Those couples who wish to avoid having an affected child can be offered prenatal diagnosis.

Primary health care services provide screening through the family doctor which allows individuals or couples to choose for themselves the most appropriate time to be screened (Watson et al 1991)

Finally, cascade screening concentrates on testing relatives of those already identified as CF carriers (Super et al 1992). As the siblings of CF carriers have a 50 per cent chance of being carriers and cousins a 1 in 4 chance, this can be an effective method of screening.

2.3.2 Prenatal screening and diagnosis

Two to five per cent of babies are born with a genetic disorder or a condition where there is a substantial genetic influence (Pembrey 1987). Figure 2.3 shows the relative frequency of those which are common and serious (Cuckle 1992).

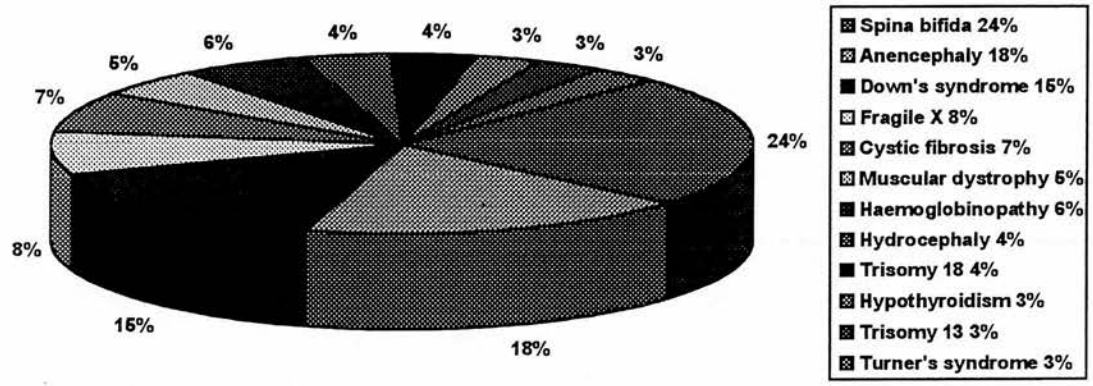


Figure 2.3 The relative frequency at birth of common serious fetal abnormalities (adapted from Cuckle 1992

A prenatal screening test aims to identify among pregnant women, who are collectively at low risk of a particular fetal abnormality, those at a higher risk. Further prenatal diagnostic tests are then offered to those who screen positive, to confirm if the fetus has a particular disorder (Donnai 1992). In the past twenty years technological developments have provided new techniques which can diagnose many fetal abnormalities. These diagnostic tools provide prospective parents with information about the risks of bearing child with a particular congenital or genetic disorder. Women come to have prenatal diagnosis via one of two routes. They may be identified as being at risk of fetal abnormality through a screening programme (i.e. a testing programme directed at the pregnant population as a whole). Or a woman may be deemed to be at risk because she has an affected child; has a family history or is a known carrier for a genetic disorder; or she may be of advanced maternal age.

The most widely used prenatal screening procedure is the ultrasound scan. which can estimate gestational age and identify multiple pregnancies. It can be used as a diagnostic tool to detect structural abnormalities in the fetus (Crespigny and Dredge 1991). It is frequently used in conjunction with diagnostic procedures namely amniocentesis or chorionic villus sampling already described (page 18).

Screening may commence prior to conception with a comprehensive serum screen (toxoplasmosis, rubella, cytomegalovirus, herpes virus) if appropriate. ABO blood group and rhesus factor can also be determined. Genetic carrier screening for haemoglobinopathies, CF and Tay-Sachs disease can be carried out. Carrier status for genetic conditions where there is an established family history for example: Fragile X syndrome, Huntington's disease, polyposis coli, muscular dystrophy (Becker, Duchenne or myotonic), neurofibromatosis and haemophilia A and B can also be offered.

At the first antenatal clinic visit a blood sample is taken for full blood count, ABO and rhesus grouping, determination of rubella state (if not already known) and serology for syphilis. Testing for toxoplasmosis is carried out, if indicated. Screening for hepatitis B is routine in some areas and it seems that HIV state may also be routinely offered in high risk areas in future (Bull 1990; Goldberg and Johnstone 1993). A urine sample is taken to exclude diabetes or nephropathy and asymptomatic bacteriuria. Urine testing is routine at every antenatal examination and blood samples are taken at regular intervals to exclude iron deficiency or other anaemia (table 2.2).

There are a number of optional screening tests during pregnancy. Ultrasound scan may be offered at 18-20 weeks gestation to exclude fetal anomalies (cardiac, renal, limb reduction defects). Amniocentesis should be offered to women over the age of 35 years or with a history of chromosome or gene defects.

Maternal serum alpha-fetoprotein (MSAFP) screening consists of taking a blood specimen at about 16 to 18 weeks of pregnancy and biochemical serum estimations of a number of markers allows detection of about 60 per cent of Down's syndrome pregnancies and approximately 85 per cent of cases of open spina bifida and effectively all cases of anencephaly. Alpha-fetoprotein (AFP) is a glycoprotein synthesised in the fetal liver and gastro-intestinal tract. It is found in amniotic fluid and in smaller amounts in maternal blood which it enters through the placenta. There is an association of an abnormally increased amount of AFP in maternal serum in a variety of fetal disorders.

An elevated maternal serum AFP (MSAFP) can indicate an open neural tube defect or an abdominal wall defect in the fetus. Abnormally low MSAFP levels are associated with fetal chromosomal abnormalities such as Down syndrome. About 4-5 per cent of women have high initial MSAFP values of whom about half will continue to have a high value on repetition of the test and will require ultrasonography. Because MSAFP levels must be calculated precisely with gestational age and number of fetuses, ultrasonography will provide the reason for raised MSAFP because of incorrect dating of last menstrual period or multiple fetuses. Around 1-2 per cent of women will require an amniocentesis to assess AFP concentration in amniotic fluid and about 1 in 10 will ultimately be found to have a fetus with a neural tube defect. At the other end of the curve another 5 per cent of all women screened will have an abnormally low AFP level. Ultrasonography will eliminate half of these women from further evaluation because of inaccurate gestational dating and the other half will be offered amniocentesis for fetal karyotyping of whom around 1 in 40 will have a chromosomally abnormal fetus. (Cunningham and Gilstrap 1991). The tests which are available during pregnancy are summarised by gestational age in table 2.2.

Table 2.2 Screening before and during pregnancy
(adapted from Bull 1990)

Stage	Urine	Blood	Chorionic villi	Amniotic fluid	Ultra sonograhy
Pre-pregnancy	Diabetes Nephropathy	Rhesus state Rubella state Haemoglobin-opathy Gene carrier status			
8-10 weeks	Bacteriuria	Syphilis Hepatitis	Karyotype Gene defects		
16-18 weeks		Anaemia Rh/ABO antibodies Alpha-fetoprotein		Karyotype Acetylcholin-esterase Alpha-fetoprotein	Multiple pregnancy Gestational age Macrosomal defects
28-30 weeks		Anaemia Rh/ABO antibodies Gestational diabetes		Haemolytic disease	Intrauterine growth retardation Placental site
34-36 weeks	Pre-eclamptic toxaemia	Pre-eclamptic toxaemia Anaemia Rh/ABO antibodies		Haemolytic disease	Intrauterine growth retardation

2.3.3 Planning a genetic screening programme

There are numerous sets of criteria published for determining the benefits of a genetic screening programme (Cuckle and Wald 1984; Holland and Stewart 1990; Simpson 1991). The disorder should be well defined and considered an important health problem. Important means that either it is prevalent in the population or that it is a serious problem. Cystic fibrosis fulfils both criteria.

The actual screening test must meet certain criteria (Cohen 1984). The test should be simple and rapid and use easily available specimens. The CF carrier test involves a simple mouth-wash of 10 millilitres of water from which sufficient buccal cells are obtained to provide DNA analysis. Carrier test results are available in one week. Ideally the test should be highly sensitive and highly specific, thus having a low percentage of false positive and false negative results.

The prenatal screening trial invited women to be screened for five common CF alleles representing 85 per cent of mutations, thus, the test failed to account for 15 per cent of CF carriers. This meant that a negative test result did not guarantee that an individual was not a carrier but reduced their risk from 1 in 25 to 1 in 167.

There must also be benefits resulting from screening. For genetic disorders, like CF, genetic counselling and either reproductive decision making or prenatal diagnosis are considered acceptable screening benefits.

The infrastructure of any screening programme requires planning and organisation. Screening and testing facilities must be available along with appropriate education of both professionals and the lay target population. Pre-screening information and counselling should provide the target population with knowledge and understanding of the risks, as well as the benefits of screening. In addition, provision for confidentiality of records and informed consent must be made, as well as counselling, diagnostic referral and follow-up of positive cases (Modell 1990). The requirements of a genetic screening programme infrastructure are summarised in table 2.3.

Table 2.3 Infrastructure required for a genetic screening programme
(adapted from Modell 1990)

- Information and education of health professionals and the target population
- System for collecting samples and delivering them to the laboratory
- Diagnostic laboratory and quality control system
- System for notifying and storing results
- Information storage and retrieval system
- An information and counselling service for carriers
- Expert counselling for at-risk couples and provision of prenatal diagnosis
- A system for monitoring the service

2.3.4 Nurses and Genetic Screening

During the 1970's there was a shift in emphasis from 'content oriented genetic counselling' which concentrated on the medical aspects of genetic disease and risk estimation, to 'person oriented counselling' with a greater emphasis placed upon the psychological aspects of genetic counselling (Emery and Pullen 1984). This move marked a steady increase in the number of nurses recruited into clinical genetic services. Nurses were considered to be ideally suited to collecting relevant pedigree and medical details and initiating preliminary investigations prior to counselling. Nurses could use their skills to assess the extent of social or psychological problems, physical or mental handicap, or anticipate language difficulties among prospective counselees. They could also assess individual knowledge, understanding and attitude toward a genetic condition as well as individual perceptions and objectives in seeking genetic counselling. Nurses could provide bereavement counselling and assess the state of grieving prior to and after counselling

The genetic nurse's overall aim was to ensure that the clinic consultation was geared to the particular needs of the individual.

In addition the genetic nurse provided clinical assistance during examination and diagnostic procedures and by being present during the counselling session facilitated assessment of individual comprehension and impact and follow-up requirements. Post counselling support of individuals and liaison with other health care professionals is now considered an important role of the genetic nurse (Guilbert 1988).

Health visitors have been a popular choice for genetic nurse specialist positions of which there are currently around 100 posts in the UK (Genetic Nurses and Social Workers Association 1994). It has been suggested that the role of the genetic nurse be reserved in genetic screening programmes to the follow up of those who receive a carrier result (Ellis 1991). This proposal stems from a concern about whether the current number of health care professionals in genetics can handle the increase of work that CF carrier screening could potentially generate. Similar misgivings have been voiced in the United States (United States Office of Technology 1992). An additional concern, in Britain, revolves around what has been described as a serious deficiency of teaching of clinical genetics in medical schools such that medical geneticists are reluctant to rely on primary care doctors to provide genetic advice (Johnston 1990). In nursing this worry also prevails and is reflected in an article devoted to genetic screening for sickle cell disease, in which midwives and health visitors are encouraged to understand the implications of the disorder and to empathise with couples at risk, but to refer to a sickle cell counsellor (Stirk 1991).

There is evidence to support these concerns from a survey of health visitors which showed that generic health visitors (those not working in genetics) have a reasonable

knowledge of the more obvious aspects of genetic services but their general awareness and perception of clinical genetic services was poor (Guilbert and Cheater 1990). Many felt their knowledge of genetics was inadequate, a factor thought to account for their failure to recognise that they themselves could initiate referral of clients for genetic counselling or screening.

The success of most preventive forms of health care depends upon public awareness, and understanding and acceptance obtained through informed discussion and consent (Rosenstock et al 1975). It is now recognised that this area is one in which the midwife, family planning clinic nurse and health visitor have a primary role and that medical genetics should be included in their core curriculum (Royal College of Physicians 1989). A community genetic counselling course for primary health workers is now available in London (Anionwu 1991). More recently, an MSc Course in Genetic Counselling has been initiated at Manchester University. Students are taught case work in human and clinical genetics, and counselling, with fieldwork in community placements and genetic clinics (Manchester University 1993). Currently, the Royal College of Physicians is carrying out an enquiry into counselling for genetic disorders with the aim of auditing the counselling that people at risk of genetic disease receive from health professionals other than clinical geneticists. The Royal College of Midwives is supporting this enquiry which is investigating specific cases where individuals are identified with one of seven genetic conditions namely: cystic fibrosis, familial adenomatous polyposis, multiple endocrine neoplasia type 2A, Down's syndrome, haemophilia, thalassaemia and neural tube defect. (Royal College Physicians 1993).

2.4 A Prenatal trial of cystic fibrosis carrier screening

The prenatal CF carrier screening trial was undertaken by the University of Edinburgh Human Genetics Unit. It was conducted through the antenatal clinics of a large Edinburgh maternity hospital, the Simpson Memorial Maternity Pavilion.

In addition to the researcher, a genetic nurse was appointed to carry out the day-to-day running of the trial working alongside the midwives in the antenatal clinic. A midwife with appropriate clinical and counselling experience, she was also responsible for the major genetic counselling component of the trial. It was felt that the researcher was better to be an outsider rather than a participant group member. Firstly, it was thought that women and their partners could become confused between the genetic nurse and the researcher. Secondly, within the antenatal clinic the staff had expectations of the genetic nurse regarding her contribution to the work. Finally, it is considered difficult for a researcher to be immersed in a work role and maintain objectivity (Field and Morse 1990).

2.4.1 An outline of the screening procedure

The principal steps in the screening procedure were as follows: a) to offer pregnant women attending the antenatal booking clinic a CF carrier test, b) to offer the partners of women who tested positive for the CF gene a CF carrier test, c) to offer prenatal diagnosis to carrier couples (Brock 1990b). The major questions to be addressed during the trial are detailed in table 2.4.

Table 2.4 The major questions to be asked in the prenatal CF carrier screening trial

1 Is it possible to organise antenatal clinics in such a way as to offer and deliver heterozygote testing to a high proportion of pregnant women?

2 Can heterozygote testing be delivered to a high proportion of the partners of women who test positive?

3 Can effective methods of counselling be established so that pregnant women and their partners have a full understanding of what is involved on entering the programme?

4 Is this form of prenatal screening acceptable to women and their partners?

5 To what extent is the programme invalidated by the incomplete nature of heterozygote testing?

6 What are the costs of the different parts of the programme?

7 How can this form of prenatal screening be integrated into National Health Service antenatal care?

Source: Brock (1990b) Unpublished research proposal submitted to the Cystic Fibrosis Trust

It was question 4: 'Is this form of prenatal screening acceptable to women and their partners' which formed the initial concept for this study. Before any initiative could be taken to answer this question the researcher reviewed in detail the discrete stages of the CF carrier screening process.

2.4.2 Organisation of the screening trial

2.4.2.1 The trial setting.

The trial was carried out in the antenatal clinic of the Simpson Memorial Maternity Pavilion, Edinburgh which has approximately 5,000 deliveries a year. An estimated 1,000 of these women booked at one of 6 peripheral antenatal clinics and were not included in the trial. The screening trial was introduced in October 1990 in one antenatal clinic and gradually expanded to run in all nine weekly antenatal clinics held in the hospital. An outline of the principal stages of the screening protocol is shown in table 2.5.

Table 2.5. Outline of the principal stages of the screening protocol

1. Invite participation with booking appointment letter
2. At clinic, collect signed consent form
3. Mouthwash sample from woman
4. If negative for mutant alleles, no further action. If positive invite couple for counselling session. Mouthwash sample from partner
5. If partner negative, counselling but no further action
6. If partner positive, refer to obstetrician

Women were offered testing by means of a leaflet sent with their booking clinic appointment (see appendix). They were asked to discuss it with their partners and were invited to join the trial by signing a consent form. Women were advised not to enter the trial if they were more than 18 weeks' gestation of pregnancy or if they could

not identify the baby's father. Other reasons for exclusion are shown in figure 2.5 page 46.

2.4.2.2 The role of the midwife in the screening trial

The midwifery team in the antenatal clinic consisted of a sister, a specialist midwife in prenatal diagnosis, and six staff-midwives. Education of clinic staff was initiated by a lecture and an educational package containing the patient information leaflet, a leaflet devoted to the disease CF, a hand-out describing the CF carrier test and outlining the delivery of the CF trial in the antenatal clinic.

The midwife's role is concerned with all aspects of antenatal care: clinical, educational and advisory. Midwives are qualified to assess the health of the mother and baby and to recognise those signs of abnormality in either which necessitate referral to medical staff for advice or treatment. (Robinson et al 1983). Midwifery training prepares midwives to advise women individually on matters such as health care during pregnancy and preparation for after delivery. They are ideally placed to recognise the emotional needs of individual women and develop a supportive and continuing relationship with them during their pregnancy at the same time as monitoring and assessing their physical well-being and that of the fetus (Sweet 1988).

In the study setting, midwives worked with doctors in obstetrician led antenatal clinics. A midwife responsible for booking a woman took a general medical history, gave general advice on diet, welfare benefits, antenatal and parent craft classes, and details of future visits. The midwife discussed routine prenatal screening tests and optional screening tests such as maternal-serum alpha-fetoprotein

and CF carrier screening. An ultrasound scan was performed to estimate gestational age and a venous blood sample taken (table 2.2). Cervical cytology was recommended and carried out if appropriate.

During the prenatal history taking the midwife obtained socio-demographic details including ethnic background, occupation and religious beliefs of both partners. She discussed a woman's or couple's expectations concerning the pregnancy and the care they hoped to receive. This information provided insight into the stability of a woman's relationship with her partner and the significance of the pregnancy to them both. Other details relevant to the CF carrier screening trial were a couple's ethnic origins in relation to gene frequency, availability of the male partner, and a couple's attitude toward fetal abnormality and termination of pregnancy.

Women who were greater than 18 weeks' gestational age, as estimated by ultrasound scan, were advised not to enter the CF screening trial because of the emotional implications of termination of pregnancy late in the second trimester (Donnai 1981; Lloyd and Lawrence 1985; Iles 1989). Rather, they were advised to be screened after the delivery of their baby and before a subsequent pregnancy.

The maternity hospital in which this study took place was situated within a health board area which had developed a shared record system. Under this scheme one copy of a specially designed antenatal card was held by the hospital in the patient's antenatal records, another by her general practitioner and both copies were kept up to date from a master record held by the woman and presented by her at each clinic visit. On the rear of the woman's liaison card the midwife responsible for booking her recorded whether or not she wished to participate in the maternal serum alpha-fetoprotein screening programme. A study concerning the attitude of general

practitioners to screening for genetic diseases revealed that not all regarded the management of tested patients to be the sole responsibility of the genetic services. Thirty per cent of general practitioners stated they wished to carry out post-test counselling support and a further 11 per cent wished to disclose the test result (Mennie et al 1990). Provision was, therefore, made to inform GPs of their patient's decision to accept or decline CF carrier screening by recording it on a woman's antenatal liaison card. Before the trial commenced, GPs were informed of the objectives of the trial in their monthly newsletter circulated to each practice.

2.4.2.3 The specialist midwife in prenatal diagnosis

A specialist midwife was responsible for giving information about prenatal diagnostic procedures. The growth of prenatal screening and diagnosis has created a specialist role for the midwife (Whelton 1989). The specialist midwife provided care, advice and support for mothers and couples considering or undergoing prenatal diagnostic tests. Her role was also one of liaison with the midwives working in the antenatal clinic and the medical staff.

Prenatal diagnosis is a screening facility for any pregnant mother at risk of delivering a baby with an abnormality. Thus women with a raised or lowered maternal-serum alpha-fetoprotein screening result, or of advanced maternal age, or with a known risk of fetal abnormality by virtue of a previous pregnancy or family history come under the care of the specialist midwife.

Fetal medicine is the management offered upon detection of problems which, with therapy, may result in a successful pregnancy outcome; for example rhesus isoimmunization and fetal urinary obstruction. The specialist midwife will care for

mothers during fetal operative procedures and give the necessary counselling and support to enable them to cope with the procedure.

In addition the specialist midwife's role is that of educator and counsellor. The invasive prenatal diagnostic procedures of amniocentesis and chorionic villus sampling require explanation to couples about the procedure itself, the risks and the significance of a positive and negative fetal test result. The counselling role revolves around the decision making process of whether such screening is a practical and acceptable course of management for a pregnancy. The dilemma of whether a couple decide to terminate or continue an affected pregnancy is frequently a counselling role undertaken by the specialist midwife as well as bereavement counselling.

The specialist midwife's role was to liaise with the antenatal clinic and the laboratories where fetal diagnostic tests were carried out and communicate results to the obstetrician, general practitioner and patient. In conjunction with the obstetrician the clinical specialist midwife undertook counselling and arranged for further investigations if deemed necessary. Being conversant with the clinical significance of genetic screening tests, the specialist midwife was frequently involved in explaining these tests to women and their partners.

Close liaison with the specialist midwife was established early on in the CF screening trial. Couples where both partners proved to be CF carriers were introduced to the specialist midwife who gave more detailed information and counselling about diagnosing CF in the fetus by chorionic villus sampling or by amniocentesis. The specialist midwife liaised with the genetic nurse to ensure that those patients undergoing prenatal diagnosis for conditions other than CF, were also offered CF carrier screening, and that those who wished their CF carrier status determined were promptly screened.

2.4.2.4 The role of the genetic nurse

The genetic nurse was on duty at each antenatal booking clinic to discuss concerns with both women and antenatal clinic staff. Studies have shown that women are not always aware of what antenatal tests they have undergone or what the results mean, often because of an insufficiency of information provided by obstetricians and midwives (Marteau et al 1992c). The genetic nurse was available to reinforce information which set out the options and factors involved in CF screening and to encourage informed decision making. Using a felt board as a visual aid the genetic nurse was able to explain genetic concepts, in a simple and non-threatening way, to those who experienced difficulty in comprehending the information leaflet. Moreover, she was available to identify and respond to staff difficulties.

Counselling carriers and their partners

In a number of cases the genetic nurse had already met a woman or couple at the antenatal booking clinic; for some it was their first face-to-face meeting with her. The overall objective of counselling was: to provide the couple with information about the meaning of a woman's positive CF carrier test result; to explore the medical and genetic aspects of CF; to prepare the couple factually and emotionally for the period awaiting the male partner's test result; and to explore the social and emotional impact of having received a positive CF test result.

During the first minutes of the counselling session the genetic nurse learned the details of the couple's experience of having received the woman's positive carrier test result. This enabled the genetic nurse to become aware of the emotional impact on the couple. Such knowledge helped judge a couple's ability to assimilate information which may have to be repeated or stressed to ensure comprehension. A number of important facts were emphasised during the initial stages of the counselling session. These were that the test

result was not a fetal indicator and that being a single gene carrier was of no great significance unless both partners were carriers. It was also stressed that all individuals carried a number of aberrant genes. A felt board was used as a visual aid to explain the possible consequence of the fetus being a single gene carrier if one parent was a CF carrier, and the significance if both partners were shown to be carriers. The availability of prenatal diagnosis was highlighted at this juncture. Detailed information about fetal diagnostic tests were discussed only if the couple wished; some couples regarded this information as inappropriate at this early stage of the screening process.

The 50 per cent chance of siblings of a carrier also being carriers was explained. However, supplying this information to relevant third parties was left to the couple. Details of carrier testing for relatives was outlined in a patient information leaflet issued to all couples.

Regarding information about the disease CF, the prevailing approach was to tailor information to meet the needs of individual couples. Explicit details were requested by some; conversely, others deemed this information unnecessary, preferring that these particulars be given if both were identified as carriers. A leaflet describing the disease was issued to all couples which granted them freedom to confront or ignore the information.

It was emphasised that a negative carrier test result was not a guarantee against being a carrier. The false negative rate of the test was explained and male partners were asked to sign a consent form before giving a mouthwash sample for DNA analysis. Helping to make the period of days awaiting the male partner's test result more tolerable was an important role of the genetic nurse. A reassurance leaflet was issued to all carriers. Information included a contact telephone number and all couples were invited to

telephone if they had questions, anxieties or simply needed to talk. Couples were told that they would be contacted by telephone within 1 week to report the male partner's test result. Alternative arrangements were made with couples who did not have a telephone. Test results were reported in writing to both partners and to their respective GPs as well as to the consultant obstetrician. All counselling information was documented in a patient information leaflet issued to all couples.

2.4.2.5 Pre-screening information

When pre-screening information and counselling is structured around a single session, as is the case with prenatal CF carrier screening, the counselling component may be attenuated. The accuracy of information about the purpose and procedure for testing are of paramount importance in helping women make an informed decision about screening. What is perhaps less obvious is that for the process to be effective, as well as accurate, counselling must take place. The counselling process is one that should not be forced, skimped or hurried (Kelly 1977) and herein may lie a problem. In a busy antenatal booking clinic there is limited opportunity for lengthy discussion. Moreover, extension of the average booking time of each patient would create mayhem in a clinical area already considered to be suffering from an excessive workload resulting in public dissatisfaction (Scottish Home and Health Department 1983). Providing women and their partners with adequate information which would enable them to make a decision regarding genetic carrier testing for CF provided a challenge.

The concept of a study to assess the attitude of the target population to a draft information leaflet, originated from research into designing protocols for breast feeding (Houston and Field 1988). Findings showed that only too frequently educational protocols were developed without seeking the reaction of the target population. It was

felt that for information and education to be effective, women should be consulted to determine what they thought they needed to know.

A draft leaflet outlining the aims of the test and describing the screening procedure was sent to 200 women along with their booking clinic appointment. Twenty women were not eligible for CF carrier screening for reasons of late gestation (greater than 18 weeks), abnormality of pregnancy (for example, blighted ovum), or unavailability of their partner. Questionnaires were issued to all remaining 180 women along with a stamped addressed envelope. The women were asked to complete the questionnaire and return it. They were not asked to identify themselves because it was thought they would feel free to comment honestly if they knew their identity was concealed. A total of 145 (81%) returned questionnaires of whom 135 were screened and 10 declined to be screened. Details of this study have been published (Mennie et al 1992a). On the basis of this study a printed leaflet was designed describing the aims of prenatal CF carrier testing and outlining the screening procedure (see appendix).

The printed leaflet was sent to all women along with their booking clinic appointment; they were asked to discuss it with their partner and were invited to join the trial by signing a consent form which was incorporated in the leaflet. At the clinic the midwife responsible for booking a woman asked if she had read the leaflet, understood it and wished to join the trial. Women who had not read the leaflet either because of visual or reading difficulties, or who found it too complex were counselled by the genetic nurse who was separate from the nurse researcher. Signed consent forms were filed in the woman's antenatal records.

2.4.2.6 Sampling and laboratory analysis

Prior to each clinic a sample form was placed in a woman's antenatal records. A patient identity label was secured to the form and details of the patient's partner and general practitioner obtained. Participants were asked to rinse out their mouths briefly with 10 mls of tap water which was transferred into a universal container. Acceptance or refusal of the test was recorded on the patient's antenatal liaison card. Mouthwash samples along with the sample forms were posted to the laboratory for analysis as described by Shrimpton et al 1991 and Ferrie et al 1991.

Patients were told that initial testing would take 7 days and that at that time they could assume that their test was negative. The leaflet intimated to women that they could receive their negative test result if they brought a stamped addressed envelope to the clinic. Only 1 per cent of women made use of this facility to receive their negative test result a majority requesting confirmation of their negative result at a subsequent clinic visit. Women with a poor obstetric history, a history of psychological disturbance or who appeared, to the midwife, to be anxious were informed by the genetic nurse of their negative test result by telephone call or letter. Specimen forms reporting the test result were returned from the laboratory to the hospital and filed in the antenatal records. The laboratory were responsible for recording all samples tested, along with their result, on a dedicated computer.

Women who tested positive were informed by telephone, if possible, or by letter otherwise. An appointment was made for counselling at the hospital, together with their partner. Counselling was carried out by the genetic nurse. The couple were given two additional leaflets, a reassurance leaflet and an information leaflet which reiterated the counselling information and outlined the screening procedure for relatives. A contact telephone number was clearly advertised on the front of the

leaflets and couples were encouraged to call if they had any concerns while awaiting the partner's test result. Partners who wished to be screened signed a consent form which was filed in the antenatal records. Partners' samples were tested as quickly as possible (average 4 days) and the results communicated by telephone. Results were recorded on the partner's specimen form which was filed in the carrier's antenatal records. Carrier and partner test results were communicated by letter to the woman and her partner, to the general practitioner (GP) and the consultant obstetrician.

2.4.2.7 Management of carrier couples

The protocol for the management of heterozygous couples is outlined in figure 2.4. In the event of a couple both being identified as CF carriers the consultant obstetrician and GP were contacted by telephone and letter. The couple were then contacted by telephone, and seen the same day at the hospital for counselling by the consultant obstetrician and genetic nurse. Additional counselling by a consultant paediatrician and clinical geneticist were offered in all cases. If prenatal diagnosis was requested it was carried out as quickly as possible by chorionic villus biopsy or amniocentesis. An appointment was made in advance with the couple to attend the hospital to receive the prenatal diagnosis result on the status of the fetus. Results were available in 48 hours and if the fetus was unaffected the couple were informed immediately by telephone and by letter. The result was also reported by telephone and letter to the consultant obstetrician and GP. An appointment was given to the couple for a follow-up ultrasound scan one week later to ensure the pregnancy was continuing uneventfully. A six week follow-up appointment with the genetic nurse was routinely given to all couples and an assessment interview schedule was completed at that time for both partners.

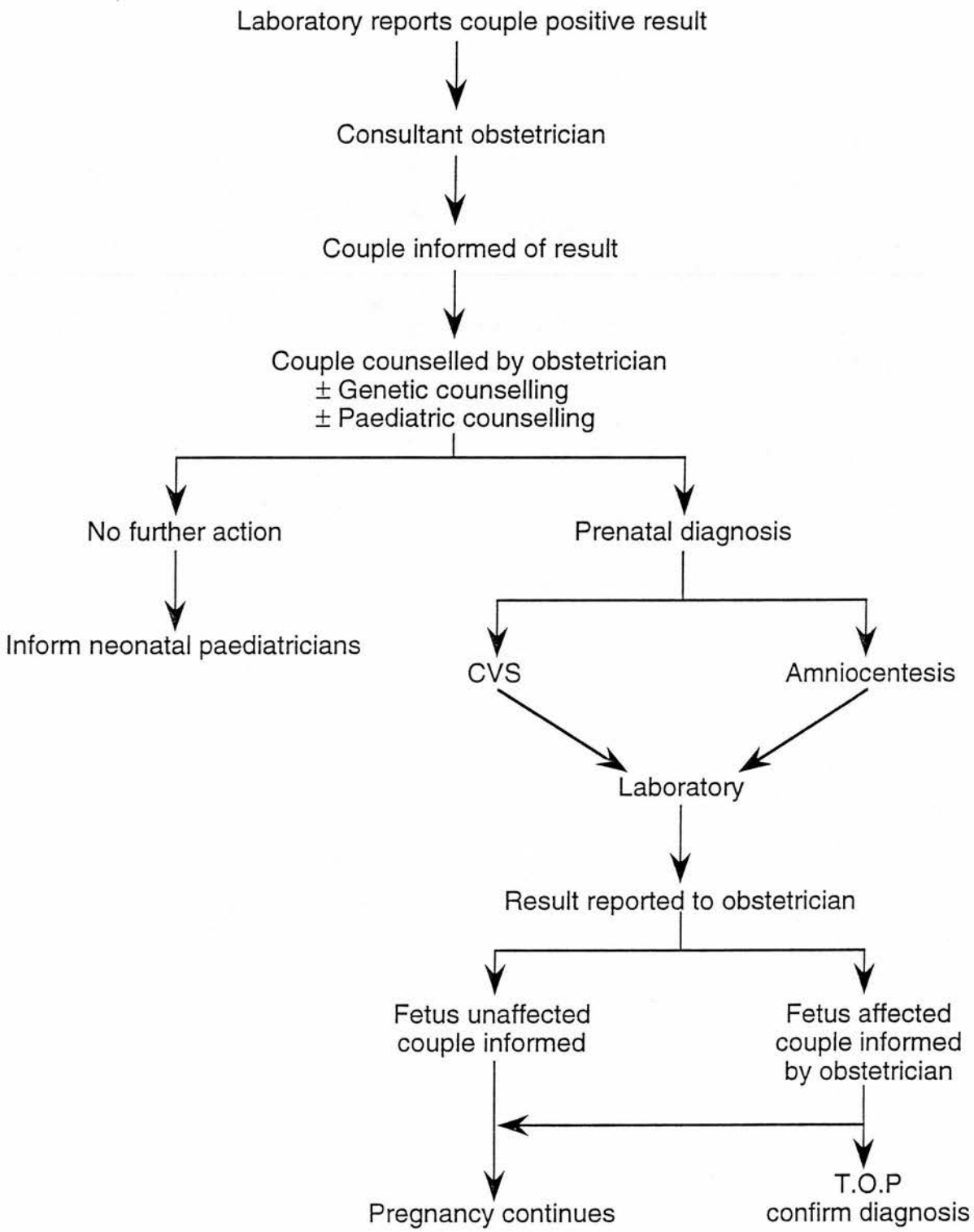
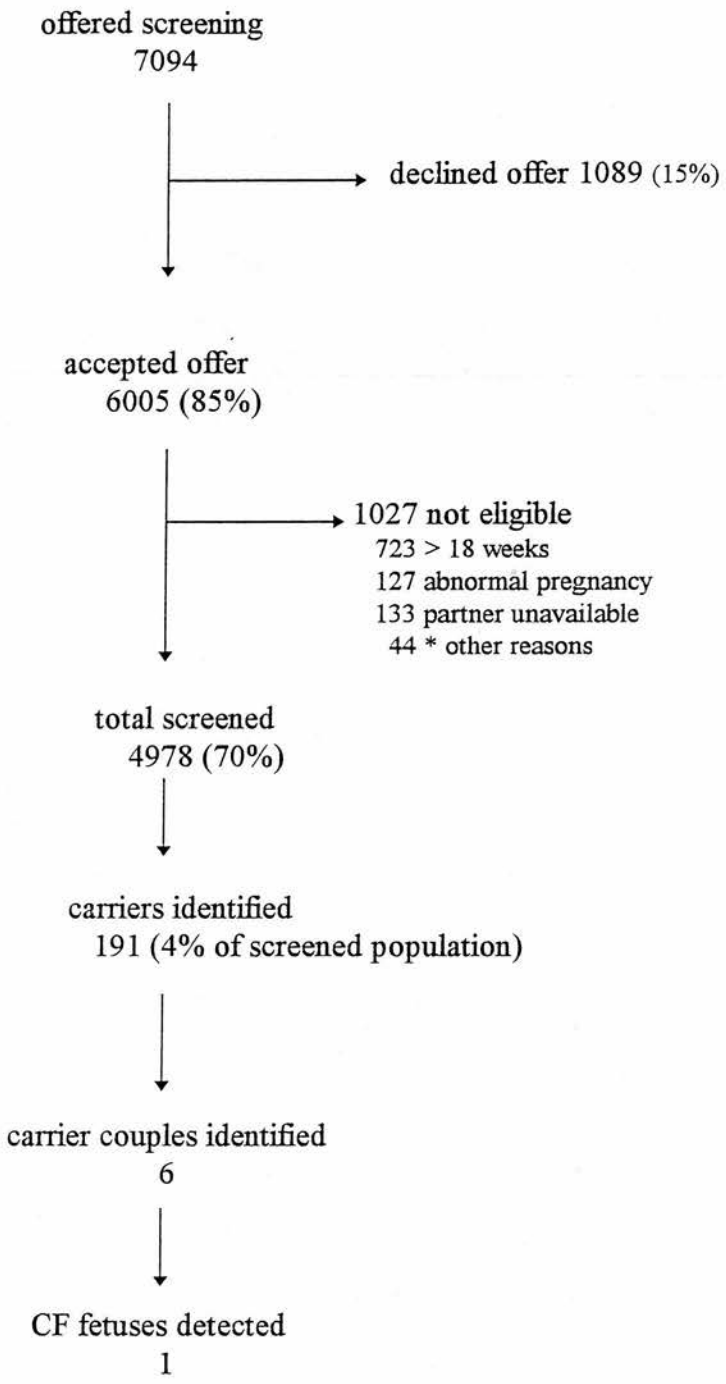


Figure 2.4 Protocol for management of heterozygous couples

In the event of the fetus being affected the GP and obstetrician were informed immediately. The couple were seen, as arranged by the consultant obstetrician and genetic nurse, for further counselling. If the couple wished to terminate the pregnancy arrangements were made to carry out the procedure at the convenience of the couple. Further discussion with a paediatrician or genetic counsellor was offered in all cases. The genetic nurse took responsibility for initial bereavement counselling, and arrangements were made for long term follow-up either through the GP or with a self-help group. All couples were given a six week follow-up appointment with their consultant obstetrician. If a couple requested termination of pregnancy to be carried out, fetal tissue was forwarded to the laboratory for confirmation of diagnosis.

2.4.3 Uptake of screening by women during the trial

From October 1990 to December 1992, a total of 7,094 women received an information leaflet inviting them to participate in the screening trial (Figure 2.5). A total of 1089 (15%) women declined the invitation to be screened. Reasons why they declined are addressed in a study reported in this thesis. A further 1027 women were not screened because they were over 18 weeks gestation of pregnancy at booking (723), because the pregnancy was not viable (127), or because they were not in contact with the father of the baby (133). Forty four women were also excluded from the trial for a variety of reasons listed in figure 2.5.



* 28 from ethnic minority groups where incidence of CF low and whose command of English was poor; 9 with low I.Q.; 1 not pregnant; 3 pregnant by artificial insemination by donor; 1 triplet pregnancy; 1 male partner screened instead; 1 HIV positive.

Figure 2.5 Uptake of screening by women during the prenatal screening trial

Among the 4,978 women tested there were 191 CF carriers identified. Partners of these 191 carriers were invited to be screened. In four cases appointments for counselling were ignored and in all cases the GP and consultant obstetrician were informed. Of the 187 partners screened 6 were found to be positive. All 6 of these couples opted for prenatal diagnosis, 5 via amniocentesis and 1 via a trans-abdominal chorionic villus biopsy.

The gestational time schedule for the carrier women from the time they were first screened to the date of prenatal diagnosis is recorded in Table 2.6. One woman was found to be carrying an affected fetus and she and her partner decided to terminate the pregnancy. The diagnosis was confirmed on fetal tissue. The other 5 women were carrying unaffected fetuses and proceeded to term and have subsequently delivered live infants. No attempt was made to monitor these infants directly but follow-up via the GP has confirmed uneventful neonatal periods in all cases.

Table 2.6 Gestational schedule for carriers from time of screening to prenatal diagnosis

Carrier	Screened	Partner screened	Partner's result	Prenatal diagnosis	Status of fetus
1	13 weeks	14 weeks	15 weeks	15 weeks	unaffected
2	10 weeks	11 weeks	12 weeks	12 weeks	affected
3	12 weeks	13 weeks	14 weeks	15 weeks	unaffected
4	13 weeks	14 weeks	15 weeks	18 weeks	unaffected
5	10 weeks	11 weeks	11 weeks	14 weeks	unaffected
6	12 weeks	13 weeks	13 weeks	16 weeks	unaffected

2.5 Some key questions relating midwifery care

Prenatal CF carrier screening aims to detect couples who are at a 1 in 4 risk of having an affected child and to offer prenatal diagnosis. To identify just one couple at risk of having a child with CF, many hundreds of women are screened. Although a majority are reassured in the long term, they have to decide whether to accept the screening test in the first place. Screening should be voluntary, but do women who are offered carrier screening perceive that they have freedom of choice to accept or decline testing?

Informed choice means being given clear accurate information tailored to suit the needs of the individuals concerned, and thus enabling them to make a fitting decision. Can pre-screening information and counselling be satisfactorily presented by midwives in a busy antenatal clinic?

Although the purpose of genetic screening is theoretically to benefit families, screening has the potential to reassure or threaten an individual (Stoate 1989). Regardless of a majority of individuals indicating that they are in favour of general health screening (Levine 1991), the public has yet to judge genetic carrier screening. Proponents of screening are alerted to the ethical obligation to ensure that the benefits of a screening test outweigh the harm (Mant and Fowler 1990) and of the necessity of monitoring the psychological stress which screening may engender (Marteau 1989a). A majority of women who undergo any prenatal screening test will receive a negative result and reassurance, but for those who receive a positive result studies which have measured their responses show that women find it is a distressing experience (Robinson et al 1984; Marteau et al 1989b).

If 5,000 women are screened for the CF gene only an estimated 8 who screened positive will have a partner who is also a CF carrier. These couples face the dilemma of prenatal diagnosis. However, an estimated 170 women are identified as single CF gene carriers and enter a period of uncertainty while their partner is screened. If the male partner's test result is negative he could still carry one of the rarer gene mutations which the test did not identify. Consequently these couples are left with a residual risk of around 1 in 640 of having an affected child, substantially higher than their starting risk of 1 in 2,500. These couples are of particular concern because doubt may be raised about the health of the fetus and residual anxieties could persist throughout the remainder of the pregnancy and perhaps even after delivery.

The question of whether offering prenatal screening generated anxiety among the pregnant population as a whole has not been addressed and if women who perceive it as stressful avoid being screened? (Marteau and Slack 1992a). For midwives the potential that the prospect of being screened creates stress among the pregnant population as a whole would raise serious questions about the advisability of offering CF carrier screening during pregnancy.

Apart from perception of the benefits or harms which can result from screening, the decision to be screened may be influenced by a person's perception of their risk of being a carrier rather than the actual risk (Marteau et al 1991a; Wertz 1984). It has been shown that when an individual is confronted with the risk of genetic disease in their offspring, they tend to be influenced more by their perceived ability to cope with an affected child than by numerical risk. Thus, their focus of concern shifts from the probability of being at risk to that of actually being at risk and the potential to bear an affected child (Lippman-Hand and Fraser 1978). In turn a woman's knowledge and perception of the effects of the disorder may also influence her decision to be screened.

A genetic screening test differs from most other prenatal screening tests in that it has a potential impact not only on the fetus but also the partner, parents, grandparents, siblings and children of the woman being screened (Elias et al 1991). With whom will women discuss the test before deciding to accept or decline the invitation to be screened?

In compliance with the philosophy of nursing and midwifery, the care of women who are offered any prenatal test should reflect their individual needs. It was clear that determining what these needs were in relation to genetic carrier testing could only be revealed by drawing on the experience of women who participated in the screening trial. The significance of such findings could reflect upon midwifery care which no longer concentrates on physical safety in childbirth but in meeting the demands of today's parents who seek from pregnancy an enriching, fulfilling experience (Sweet 1988). The previous section considered the scientific and technological advances which have already made a major impact on the care of the pregnant woman who is subject to a wide variety of investigations throughout pregnancy. An increasing number of these investigations detect abnormal conditions which could have an adverse effect on the developing fetus. It is to the midwife that women may turn for help in deciding how to use these tests wisely and selectively. But, just because care in pregnancy has become increasingly sophisticated, it does not mean that women no longer experience minor disorders of pregnancy which can cause physical and emotional discomfort and for which they also turn to the midwife for advice and comfort. Increasingly demands are being placed upon the skills and knowledge of the midwife, but, as genetic technology advances and more tests which promise to raise fundamental ethical issues become available, further demands are placed upon the attitudes of midwives. As midwifery moves from a traditional to a woman-centred approach, midwives have to strike a balance between the needs and demands of women and the major scientific and technological advances sometimes thrust upon them.

In Britain, population screening for genetic disease has been confined to the ethnic minority groups who are most at risk for the following conditions: sickle cell disease, thalassaemia and Tay-Sachs disease, thus, the concept of genetic screening is not one with which most individuals are familiar. But perhaps the greatest concern must be that technological advances may run ahead of nursing and midwifery knowledge and genetic screening for CF is already underway before the profession has had time to consider the implications for patient care and nursing commitment.

The impetus for research is its valuable role in defining and limiting problems which new procedures in prenatal care may generate (Sweet 1988). Exploring the effects that a new technology in reproduction has upon women and their partners may generate knowledge which will enable midwives to provide the best possible care to mothers, fathers and babies.

Although the researcher was separate from the genetic nurse who was responsible for the delivery of the screening test, the two worked closely together. The advantages of developing a complementary partnership between nurse practitioner and researcher has been highlighted (Tierney and Taylor 1991; Titchen and Binnie 1993). In a study designed to improve nursing practice on the basis of research, the practitioner was found to provide a clinical perspective to the research issues and questions while the researcher provided academic input. Although the researcher and practitioner had differing interests and priorities, neither found this to inhibit close collaboration (Tierney and Taylor 1991). Similar mutual benefit was experienced in an action research study to alter nursing methods in an acute medical unit from a traditional approach to a patient-centred approach (Titchen and Binnie 1993).

The collaboration between the genetic nurse and researcher in the present study quickly revealed mutual advantages. The researcher found that having a colleague who was familiar with the antenatal clinic setting, its day-to-day workings and the patient population was invaluable. The genetic nurse could advise on whether a model of research would be acceptable to pregnant women and antenatal staff. Furthermore, she was able to check the relevance and readability of educational materials designed for women and staff. The advantages to the practitioner were that she benefited from the feedback of the researcher's findings and observations and was able to integrate these into clinical practice. The added advantage of both parties sharing the same beliefs, concepts and ability to compromise cannot be overstated. Consequently it was possible to design a study which was an integral part of the delivery of the screening trial and which would contribute to patient care. The conceptual framework of the study is outlined in the next chapter.

CHAPTER 3
CONCEPTUAL FRAMEWORK

Conceptual Framework

It was apparent from an initial literature review that most women who receive a positive prenatal screening test result receive the news with some degree of distress. Whether the offer of screening itself could generate stress among the pregnant population as a whole had not been answered. This led to a review of the subject stress and coping and models of stress and coping. The conceptual framework for the study was drawn from a model of stress, coping and mental health (Cochrane 1983). The model was used as a frame of reference to help understand and interpret participants' reactions to CF carrier screening and develop the research questions.

3.1 The effect of stress on pregnancy outcome

Work to date indicates that stressful events may have a detrimental effect on the outcome of pregnancy, particularly in relation to pre-term labour (Newton et al 1979; Berkowitz and Kasl 1983). Mothers who have delivered pre-term infants have stated that they had experienced a period of stress more frequently than a control group. Moreover, compared to controls, these women expressed more negative feelings toward the pregnancy. Also of significance was a feeling of apprehension about labour, delivery and motherhood (Muylder1989). The possibility that maternal stress may have a direct effect on the fetus is an additional concern. Increased maternal arterial pressure and increased maternal muscle tone reducing intrauterine space has been demonstrated (Hepper 1989) and, it has been suggested that pregnancy anxiety is associated with increased fetal activity, as well as hyperactivity and irritability in the neonate (Field and Garcia 1985).

3.2 Stress

Stress tends to be referred to in an abstract way and needs to be defined. Stress has been defined as "a fashionable term denoting usually disagreeable stimuli; physiological, behavioural and subjective responses to these; or the whole stressful situation." Four types of stress can be recognised: 1) acute time-limited, for example, awaiting surgery, 2) sequential - one event initiating others that occur over a period, for example bereavement, 3) chronic intermittent, for example conflicts with neighbours and 4) chronic, for example, being disabled"(Wilkinson 1992 page 9).

Goldberg and Huxley (1992) propose a triad of elements which determine the sequence of events that evolve when an individual is faced with a stressful situation. The first is 'vulnerability to provoking agents' which is determined by several factors in an individual: genetic factors, experiences of parenting and experiences during childhood, personality variables, and the individual's current social situation. Thus, an event may cause a stressful reaction in one individual but not in another. The second element is 'destabilisation' which refers to the process of the individual beginning to experience symptoms, which is determined by the severity of the provoking agent and the vulnerability of the individual. Thus, the individual meaning of a stressful event will vary. Moreover, a stressful event may have a negative consequence or a positive consequence (Lennon 1989). The third element in the triad is 'restitution' which refers to the process of losing symptoms. The factors which determine how long a stressful reaction lasts are, to some extent, dependent upon vulnerability and the provoking agent, however, social factors are also crucial (Goldberg et al 1990).

Social support both before and after a crisis is a factor in restitution, as is crisis support of an ongoing and constant nature (Brown et al 1986). Stress occurring during

ongoing difficulties may cause delay in restitution; for example, physical illness, social problems such as housing, unemployment, financial difficulties, bereavement and problems with interpersonal relationships (Goldberg and Huxley 1992). Conversely the occurrence of positive life events can contribute to rapid restitution (Brown et al 1988; Brown et al 1992).

3.3 A review of models of stress and coping

3.3.1 Stress as a response.

This approach treats stress as a dependent variable. Stress is described as the individual's response to disturbing stimuli. One of the founding fathers of stress research, Hans Selye, postulated a General Adaptation Syndrome (GAS) of somatic symptoms produced by 'non-specific stress' (Seyle 1974). This was the first attempt to explain the process of stress-related illness. Seyle suggested that the body's adaptability was limited and if exposed to constant stress exhaustion would result. Seyle's GAS model is divided into three stages of stressful response. The first phase is one of alarm reaction during which the individual is in an initial shock phase of lowered resistance. This is followed by a counter-shock reaction during which the individual's defence mechanisms are activated. Severe and prolonged stress may result in the individual's resistance collapsing. The second phase is one of resistance and involves maximum adaptation during which the individual may return to a state of equilibrium. However, if the stress continues or the individual's defence is inadequate they will move onto the third phase which is one of exhaustion in which the adaptive mechanisms fail or collapse and the signs of the alarm reaction reappear. More recently researchers have challenged Seyle's theory by suggesting that the presence of stress alone does not determine these physiological responses, but rather the psychological impact of stress upon the individual (Cox 1978).

3.3.2 Stress as a stimulus

This approach views stress as an independent variable. External forces are seen as placing pressure on the individual and the response of the individual will depend upon his or her individual make-up along with the severity and duration of the pressure. Stress is seen as something which happens to an individual and not something which happens in him or her. Individual variation of response is explained in terms of specific personality attributes, early formative experiences, and inheritance (Cooper and Marshall 1978). The drawback with both these approaches to stress is that neither takes into account individual perception of a situation. Stress has to be perceived or recognised by man (Cox 1978).

3.3.3 An interactionist approach to stress.

This is now the most popular model of stress. It is able to account for individual differences both in perception and in response to stressful situations. Lazarus (1966) is one of the best known proponents of the interactionist approach to the stress phenomenon. Lazarus does not see stress simply in terms of environmental pressure but rather it depends on the perception of the individual and upon the individual's physical and cognitive coping abilities. The intensity of the stress experience depends on the degree of perceived threat. If individuals have confidence in their coping ability then they are less threatened. However, if they are unsure about their coping ability they feel defenceless and overwhelmed by the threatening situation.

The stress and coping theory of Lazarus (1966) is based on the view that stress is a relationship between the person and the environment which is appraised by the individual as exceeding his or her resources, and thus endangers well- being. Coping refers to the cognitive and behavioural efforts of the individual to manage stress. Subconsciously an individual will assess a stressful situation immediately (primary

appraisal) and evaluate it in relation to well being. Therefore an event may be appraised as irrelevant, harmless, or harmful. The individual then carries out a second appraisal during which he or she evaluates the adequacy of their resources either to prevent harm or to improve their prospect of a beneficial outcome. Thus during secondary appraisal the person evaluates their coping options. Coping is seen as a buffer which moderates the impact of stress. Lazarus (1982) proposes two methods of coping. The first he terms 'emotion focused coping', when the individual attempts to alleviate emotional distress. For example, denial can alleviate stress in a particularly stressful situation by allowing an individual to continue rather than become overwhelmed. The second method Lazarus terms, 'problem focused coping' when the individual attempts to deal with the problem causing the distress. Problem focused coping is an active confronting process that gathers and uses new information to respond to a stressful event. Genetic counselling contains a major information giving component. Frequently genetic counselling takes place in the wake of stress, as a result of fetal or infant loss, loss of a child's health or loss of individual health. Both emotion focused and problem focused coping takes place as individuals strive to understand the cause of their loss and come to terms with it.

3.3.4 Cochrane's model of stress and coping

Cochrane proposes that an individual's response to a stressful event will be influenced by pre-existing vulnerability, personal and social resources and the availability of alternative responses (Cochrane 1983). Thus the immediate stressor may occur in the context of other recent events (bereavement, unemployment) which if not yet resolved may make the immediate stressor particularly threatening. The individual is already at a high level of arousal and may already be anxious or depressed. In addition past coping experience and the individual's perception of their own coping ability will influence their response. But, if

they are already attempting to cope an individual could quickly become exhausted of those resources which would allow them to deal effectively with the new stressful episode. The intervening variables described by Cochrane which may affect both the response and outcome to a provoking event are illustrated (Figure 3.1). The value of Cochrane's model of stress and coping to the researcher is that it details those factors which should be taken into consideration in measuring reaction to stress.

The premise of Cochrane's model that individual response to stress does depend upon the person's perception of how threatening a situation is, but in addition, must take into account concurrent stress, was considered an appropriate conceptual model for helping to understand how a pregnant woman might respond to CF carrier screening. The immediate stressor, receiving a positive CF test result, could occur in the context of other recent events (symptoms of early pregnancy, recent bereavement, poor obstetric history, unemployment, to name only a few possibilities). If not successfully resolved this could make CF carrier screening particularly threatening. This was considered highly relevant to the present study because pregnancy itself is recognised as a time of psychological upheaval (Blumberg 1984). If a woman felt she was coping satisfactorily with the physiological and psychological changes associated with pregnancy this would contribute favourably to her self esteem and attitude to pregnancy and, in turn, contribute to her emotional well being and ability to cope with a positive screening test result. If, however, she were to experience quite the opposite and in addition receive a positive CF test result this could affect her ability to cope.

Cochrane acknowledges that there are individual and biological differences in the extent to which an individual is vulnerable to stress but that the impact of other factors (namely sex, age, social class, and environmental factors such as housing, unemployment and

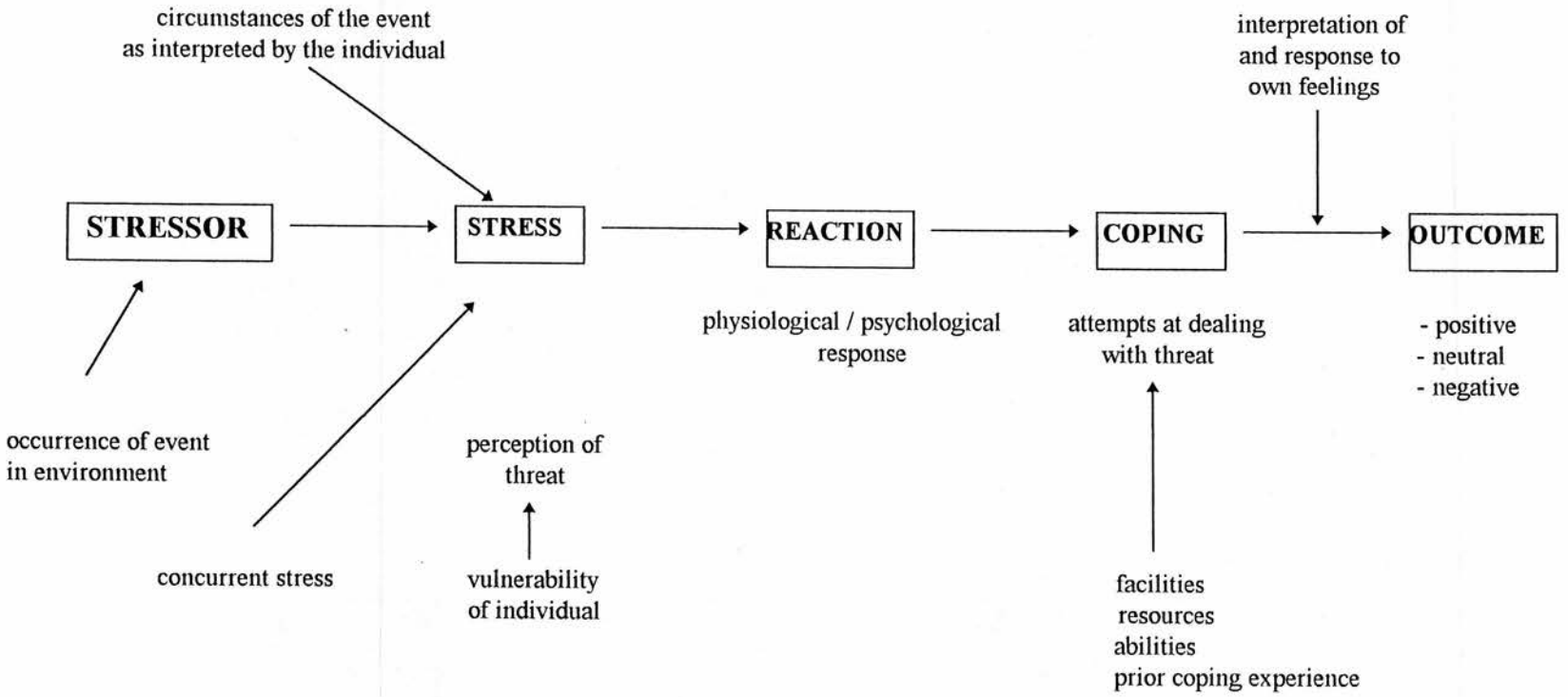


Figure 3.1 A model of stress, coping and outcome (adapted from Cochrane 1985)

social support) are important. This is supported by the results of numerous studies (Cobb 1976; Bebbington et al 1981; Cochrane and Stopes-Roe 1981; Goldberg and Huxley 1992). These factors were also considered highly relevant to the present study.

3.4 Crisis intervention

A woman who receives a positive CF carrier test result during pregnancy may experience an acute stress reaction. Such a reaction is defined by Wilkinson as the result of being exposed to " an exceptional medical or physical stressor followed by the onset, within one hour, of generalised anxiety and/or any two of the following : social withdrawal; narrowing of attention; apparent disorientation; anger or verbal aggression; despair or hopelessness; inappropriate or purposeless overactivity; and uncontrollable and excessive grief." (Wilkinson 1992, p10). The author advocates supportive treatment and gives the prognosis as favourable.

Receiving a positive CF test result during pregnancy may be regarded as a potential situational crisis. For some individuals their perception of the event, their available coping methods and available support systems may counteract a crisis (Aguilera 1990). Caplan (1964) defines crisis as a period when an individual encounters an obstacle to important life goals and failing to find a solution by usual coping mechanisms experiences upset to their emotional equilibrium. Tension occurs and is manifest by symptoms of anxiety, fear, guilt, shame and helplessness (Caplan 1964). The individual must either find a solution to the problem or adapt to failure to solve it. Either way a new state of emotional equilibrium results which may leave the individual in a better or worse state of mental health.

Caplan also advises that crises have particular relevance for therapeutic intervention (Caplan 1964). The result of a crisis is generally not the result of preceding factors such

as the nature of the problem, the individual's personality or experience. These factors may influence the outcome but the actions of the subject and the intervention of others are more important. Situational support from those who are key figures in an individual's environment is pivotal to the process of coping. There is an increased desire to be helped by others; the individual signalling this by typically being more open and amenable to outside intervention than during periods of psychological equilibrium (Caplan 1964).

Caplan (1964) also observes the considerable impact of prevention on a favourable outcome and he specifies three levels of prevention: primary prevention may either alter stressful conditions or attempt to strengthen the individual so they can resist stress and cope in adversity; secondary prevention consists of detecting problems early before they become serious; and tertiary prevention seeks to reduce the long term consequences of stress. The aim of crisis prevention is to assist the individual maintain a state of emotional equilibrium, whereas crisis intervention assists the individual attain a state of crisis resolution and regain a state of emotional equilibrium (Aguilera 1990).

Anxiety in response to receiving a positive test result could be considered healthy. Such a response has been shown to be a positive coping response to threat and contributes to a better overall outcome. Women who experienced premature birth were observed to cope better overall if distressed at the time their infant was in most danger (Caplan 1964). Those who denied the existence of any danger fared worst. In addition, women who had a supportive partner adjusted better to the crisis.

Situational crisis can result from status or role change. Threat or danger is perceived when an unacceptable role is forced on an individual causing anticipatory grief (Aguilera 1990). A woman who receives a positive CF test result during pregnancy may perceive a threat to her role of healthy mother to disease carrying mother; of mother with a healthy

pregnancy to mother with a pregnancy at risk; ultimately she may feel her role as a mother is threatened by fetal loss. Aguilera outlines the anticipatory grief which situational crisis can engender (Aguilera 1990). In relation to CF carrier screening it could be all too easy for a woman to focus on the threat of loss of a healthy baby and to withdraw from establishing a relationship with the fetus. Concern associated with this withdrawal, coupled with the label of 'gene carrier'; her perception of failure to proceed uneventfully through pregnancy; and her possible perceived inability to cope with the whole situation could precipitate a crisis.

Aguilera (1990) also identifies the influence of the value which a woman or couple place on their role, which will reflect upon the impact of a change in role. Thus a woman's attitude to pregnancy at the outset could influence her response. The greater the conflict between a woman's expectations of her role and the change she perceives brought about by receiving a positive screening test result, the harder she may find it to cope and adapt. Thus a woman's vulnerability, the circumstances of the event as interpreted by her and her subsequent perception of the threat will influence the impact of the event (Cochrane 1983).

3.5 Using Cochrane's model of stress, coping and outcome to formulate a literature review

Using Cochrane's model a number of areas related to prenatal carrier screening which could be defined as stressors or stimuli were identified. In addition, a number of concurrent stressors were identified which could reflect on an individual's response to prenatal carrier screening. These key areas are listed (Table 3.1).

Table 3.1 Key areas reviewed in relation to prenatal genetic screening.

Primary stimuli or stressors
Genetic screening Genetic disease Prenatal screening Early infant loss
Concurrent stressors
Pregnancy Concurrent life events

From these key areas a number of questions emerged: genetic disease, genetic screening, prenatal screening and diagnosis all generate their own set of tensions. What are these elements of tension? What lessons can be learned from previous genetic and prenatal screening programmes about the effects on participants?

Early infant loss may be experienced through termination of pregnancy for fetal abnormality, or loss of infant health through diagnosis of a congenital or genetic disorder. Either may occur as a consequence of prenatal genetic screening. What is known about stress and coping in individuals who experience this loss ?

It is well recognised that pregnancy can be a time of psychological upheaval. What is known about stress in pregnancy? In addition, women may also be experiencing stress in their lives which is unconnected with pregnancy. What are these stressors likely to be?

A literature review was undertaken which explored each of these topics in relation to stress and coping. The aim was to use the findings of the review to define a series of questions which would measure and describe the impact of prenatal CF carrier screening among pregnant women. The findings of the literature review are presented in the following chapter.

CHAPTER 4
LITERATURE REVIEW

The literature review detailed in this chapter focuses on those areas listed in table 3.1 of the previous chapter and considers the effects of each circumstance on an individual or couple.

4.1 Genetic screening

In the 1970's carrier screening became possible for a number of genetic disorders namely Tay-Sachs disease, and the haemoglobinopathies such as sickle cell disease, and thalassaemia. Within the United Kingdom these disorders are limited to ethnic minority groups. However, screening programmes were initiated in countries where these at-risk populations were either indigenous or had formed large communities. CF carrier screening can, at least to some degree, benefit from the lessons learned from these early genetic screening programmes.

4.1.1 Tay-Sachs disease carrier screening

Tay-Sachs disease is a lethal genetic disorder which is inherited, like CF, in an autosomal recessive fashion. It affects the nervous system causing mental retardation, blindness, seizures and ultimately producing a vegetative state with the child dying at 2 to 4 years of age (Gelehrter and Collins 1990). Predominantly Jews of Eastern and Central European descent (Ashkenazi Jews) are affected, among whom about 1 in 30 are single gene carriers, although it does occur infrequently in the general population.

In the 1970's by measuring the deficient enzyme in Tay-Sachs disease, hexosaminidase A, and later in the 1980's when the gene was identified, by DNA based analysis, single gene carriers could be identified. The Jewish population made a concentrated effort to screen whole communities (Kaback et al 1974). In New York, matchmakers arranged

for individuals to be screened prior to making arranged marriages (Merz 1987) and in Montreal a school based approach estimated that 75 per cent of pupils were tested. Studies related to the effects of screening among pupils cited a loss of self-image among 10 per cent at the time of learning of their carrier status. However, an 8 year follow-up of these pupils found they had a positive attitude toward screening and had subsequently made use of the results (Scriver and Clow 1990; Zeesman 1984). Widespread screening among the Ashkenazi Jewish population has virtually eliminated Tay-Sachs disease in this group over the last 20 years (Gelehrter and Collins 1990). Pivotal to the success of the Tay-Sachs screening programmes was the education effort devoted not only to those screened, but also to the general public. This effort continues today through nationwide mailing to community and religious organisations, colleges, schools and libraries. Despite this, a study revealed that the trend in Tay-Sachs screening suggests that both the public and health care professionals perceive genetic carrier testing as part of prenatal, rather than pre-pregnancy care (Clarke et al 1989; Shapiro and Shapiro 1989).

A group of 404 physicians of whom 231 were obstetricians and 173 were family physicians or internists (junior hospital doctors) in the New York State Health Services Area, where a Tay-Sachs carrier detection screening programme had been in operation for the past 10 years, were sent a questionnaire. The questions were designed to ascertain whether or not subjects referred patients for Tay-Sachs carrier screening; and why they tended not to refer them; to assess the frequency and objective of each referral; to explore reasons why referral was delayed until pregnancy; and to determine whether doctors had a method of identifying those who should be screened. A majority of obstetricians (77%) referred patients for Tay-Sachs screening while a majority of internists or family physicians (89%) did not. More than 80 per cent of cases referred were pregnant women and their partners. Although 61 per cent of physicians stated they

did not deal with the target population, more than half indicated that they were unable to identify who the target population were. The authors suggest that physicians who believe they are not dealing with the target population may not actually recognise who they are. Moreover, of those non-referring physicians 21 per cent were unaware of the availability of Tay-Sachs screening and those who were aware tended to refer once a woman was pregnant. This study highlighted the need for education among both the public and health care professionals about the availability of pre-pregnancy screening and to clarify who might benefit (Shapiro and Shapiro 1989).

4.1.2 Sickle cell disease carrier screening

Sickle cell disease affects haemoglobin (Hb) in red blood cells. Haemoglobin A is found normally in red blood cells, but in sickle cell disease Hb S is found, causing the cells to become deformed (sickle shaped). The sickle shaped cells become trapped in the circulatory system resulting in poor oxygen transport which can ultimately damage organs and tissues with episodes of extreme pain in the limbs, back, abdomen and chest lasting days or weeks. There is no cure for the disease and up to 30 per cent of children die in the first years of life from bacterial infections. Sickle cell disease is also an autosomal recessive disorder with the mutation occurring in the beta globin gene on chromosome 11. Sickle cell carriers have red blood cells containing both Hb A and Hb S and are usually healthy individuals (Weatherall 1991b) although defects in urine concentration, haematuria and minimal sickling of red blood cells have been reported (Sullivan 1987).

Around 1 in 12 individuals of African descent are carriers of sickle cell disease (Gelehrter and Collins 1990). Correspondingly, it became possible in the early 1970's to identify carrier and non-carrier sickle cell individuals by screening blood samples for Hb S (Weatherall 1991b). However, while Tay-Sachs disease screening programmes were

labelled successful, sickle-cell disease programmes were referred to as "screening gone wrong" (Roberts 1990 page 18). The problem was that many states in the USA passed laws making screening of new-borns, pre-school children, pregnant women and couples applying for marriage licences compulsory. These laws were passed at the height of the American civil rights movement and were perceived as racist, and eugenic, aimed at reducing the Black population. Whereas the Jewish community had developed an infrastructure of education and counselling for Tay-Sachs screening, no such supporting system existed within the sickle cell testing programmes. As a result, confusion arose over the distinction between sickle cell disease and sickle cell carriers. Moreover, there was a singular lack of public education input which resulted in discrimination and stigmatisation of carriers through public ignorance. These shortcomings were rectified by the late 1970's but the reputation of sickle cell carrier programmes was badly damaged (Rowley 1984). Today, new-born screening programmes and prenatal screening are delivered within an infrastructure of education and counselling and mandatory privacy of screening results (United States Congress Office of Technology Assessment 1992).

4.1.3 Thalassaemia carrier screening

Beta-thalassaemia is also a haemoglobinopathy causing diminished haemoglobin and resulting in anaemia, frequent infections, splenomegaly and growth retardation. There is no cure and therapy revolves around transfusions, folic acid supplements and antibiotic therapy. Beta-thalassaemia is inherited as an autosomal recessive disorder. The mutation causing the disease also occurs in the beta globin gene but, unlike sickle cell disease, there are a large number of mutations and these determine the severity of the disease which ranges from mild to severe (Cao et al 1989).

Each ethnic group has its unique distribution of mutations. Beta-thalassaemia occurs most frequently among Mediterranean, African and Asian populations. In Cyprus 1 in 7 individuals is a carrier and in Sardinia, prior to the initiation of carrier screening, 1 in every 250 live births was affected. The programmes run in these countries warrant examination especially with regard to education. In Cyprus, the Greek Orthodox Church recognised the problems associated with the high incidence of the disease. Although reluctant to endorse termination of pregnancy, they used their influence to insist that couples presenting for blessing of marriage or engagement produce a certificate proving they had been screened and had received genetic counselling. The prevention rate of affected births in Cyprus is estimated to be 97 per cent (Angastiniotis 1990). Also of note is the reported decrease in time spent on counselling which is attributed to extensive public education programmes.

In Sardinia, screening programmes have actively pursued relatives of identified carriers and encouraged them to be screened. A mass public education programme has resulted in a decrease of affected births from 1 in 250 in 1974, to 1 in 1,200 in 1991 (Cao et al 1989). Both countries report that those few affected births are primarily due to unawareness of screening and to a much lesser extent because of ethical reasons where couples decide against abortion. With so few affected babies born, the question of freedom of choice arises. How difficult is it for a couple to continue an affected pregnancy in a society which so aggressively promotes screening?

In the United Kingdom, screening programmes for beta-thalassaemia have included treatment and prevention incorporated into primary health care. Education through schools, information posters and self-help groups has alerted relevant families, couples and communities to genetic risks and services. Fewer births have been avoided in the UK but perhaps this is not surprising given that those at risk are not

the indigenous population, but ethnic minority groups dispersed around the country many of whom find prenatal diagnosis and abortion socially and religiously unacceptable (Modell and Petrou 1988). The United States, Canada, Southeast Asia, Hong Kong, South China and East Mediterranean countries all have beta-thalassaemia screening programmes (Chan et al 1991; Loukopoulos 1991; Zhang et al 1991).

The conclusions that can be drawn from reviewing these antecedent genetic screening programmes are that participation in genetic screening programmes should be voluntary, not compulsory. This important element of a screening programme has been emphasised by many (Colten 1990; Hodgkin and Yoxen 1985; Kings Fund 1987; McGregor 1990; Sutton 1990; Weatherall 1991a). Those programmes which have relied on voluntary participation (Tay-Sachs disease and thalassaemia) have been more successful and avoided major stigmatisation problems than those (sickle cell screening) where screening was compulsory. Informed consent should, therefore, be obtained prior to screening.

Public education is necessary to avoid stigmatisation of carriers and to promote familiarity with the disease. Moreover the various options which screening can offer is limited if carried out during pregnancy as outlined in figure 2.2 page 19. Education of health care professionals is required if those who might benefit from screening are to be referred at all and pre-pregnancy screening encouraged. Consumer knowledge, not only about the availability of screening but about the disease itself, has been shown to be important. As sickle cell screening in the United States revealed, it is only too easy for carriers to see their status as a health risk. Thus it appears that consumer understanding of the disease and in addition the harmlessness of the single gene state is crucial. Moreover, parallel public understanding is needed to help avoid stigmatisation of carriers.

Perhaps the most important conclusion thus far is that information and education are essential components of any screening programme if adverse consequences of screening are to be avoided. In relation to the CF carrier prenatal screening trial, the intention was that the responsibility for much of the pre-screening information and counselling, which is a necessary prerequisite to informed consent, would rest with midwives.

4.1.4 Cystic fibrosis carrier screening

One of the main aims of those CF carrier screening trials funded by the Cystic Fibrosis Trust was to look at a variety of settings in which testing could be offered. From other screening programmes it is clear that the setting does influence both uptake and acceptance. Those programmes involving the community in the delivery of screening (Tay-Sachs disease and thalassaemia) fared better than those run by governments, being imposed and controlled by a group outwith the target population (sickle cell screening in the USA). This situation would not arise in Britain where the target population are indigenous, nonetheless, how and where screening is delivered is likely to affect public acceptability.

Two British studies prospectively assessed the attitude of the public to CF carrier screening. The first by Williamson and co-workers questioned 166 males and females from two schools, two doctors surgeries, and a family planning clinic about their knowledge of CF and attitude to carrier screening. Participants were from inner city and suburban areas and from a variety of ethnic backgrounds. Over two thirds had heard of CF. Females, Caucasians and those in the older age groups were more likely to have heard of the disease. Only 50 per cent of subjects knew it was an inherited disorder while a third knew it was serious and a quarter that it affected the lungs. Over 80 per cent of subjects expressed interest in finding out if they were CF carriers but stated a preference

for screening to be carried out through their GP. None of the participants were pregnant. (Williamson et al 1989).

A study by Cobb et al (1991) involved a random sample of school children aged 14 to 16 years who were asked to complete a series of questionnaires designed to determine their knowledge about CF and their attitude to screening. Of 216 participants 75 per cent had heard of CF but only 17 per cent knew that the condition affected the lungs. Familiarity with the disease corresponded with knowledge of an affected individual. After a short lecture 88 per cent understood what recessive genes were, 98 per cent that carriers were not affected by the disease, and 99 per cent knew that an affected individual's life span was reduced. Ninety five per cent were in favour of neonatal screening and 86 per cent were in favour of carrier screening, with 87 per cent agreeing that prenatal diagnosis should be offered to couples where both partners were CF carriers. The importance of community education prior to population carrier screening commencing is emphasised and the need for non-directive counselling support. The researchers recognised the important role that school teachers might play in promoting community education and advocated that the offer of carrier screening be made in schools (Cobb et al 1991).

A study from Belgium of 385 students in psycho social studies age 20 to 57 years of whom 78 per cent had children assessed by questionnaire their knowledge about CF and attitude to carrier screening and prenatal diagnosis. 59 per cent were familiar with the name CF, but only 38 per cent could cite at least one feature of the disease and only 12 per cent were aware that CF was caused by a gene abnormality. 63 per cent expressed interest in knowing their carrier status, but only 2 per cent would choose to be screened at the beginning of pregnancy. Approximately half indicated they would forego pregnancy if both they and their partner were carriers, with an equal number stating they would make use of prenatal diagnosis. Only 4 per cent would have opted for a pregnancy

without prenatal diagnosis. Choosing to forego pregnancy was associated with perceived burden of the condition and with age; younger participants were more likely to opt for pregnancy with or without prenatal diagnosis. The perceived burden of the condition did not correlate with socio-demographic features but did with life values. CF was perceived most burdensome by those who valued pleasure and relaxation or a healthy life. The population studied were biased toward the older age group (20% were less than 30 years of age) and only 19 per cent were contemplating a pregnancy making it difficult to draw similarities with a child bearing population. Nonetheless, this was a group of well educated individuals who as psycho social students might be expected to know more about a disease like CF than the general population would. Two findings are of interest: firstly, the correlation between life values and perceived burden of CF which may be an influencing factor among pregnant women also; secondly that younger individuals felt less disposed to foregoing pregnancy and would opt for prenatal diagnosis. The 19 per cent of the population who were said to be "still contemplating pregnancy" in all likelihood were the younger age group (Decruyenaere et al 1992).

Based on the experience of screening for thalassaemia in Italy Modell (1990) proposes that "there is no single right approach" to screening for CF carriers (page 477).

4.2 GENETIC DISEASE

The writings of early societies are evidence that for centuries people have recognised and taken a keen interest in genetic traits, particularly in relation to providing rules for choosing a spouse. Hindu sacred books dating back 2,000 years, provided instructions that when choosing a wife no heritable illness should be present and that the family should show evidence of good character for several preceding generations. Indeed the ancient Hindu caste system is based on the assumption that desirable and undesirable traits are passed from generation to generation and that an individual's worth is determined at birth.

The ancient Greeks in their poetry and philosophical literature recognised the heritable traits in humans. In Homer's Odyssey the hero Ulysses encounters the Cyclops, a giant with one eye placed in the centre of his forehead and this was recognised as inherited since the story tells of an entire race on an island where Ulysses landed. The Jews in the Talmud gave genetic advice and described the inheritance of haemophilia (Cummings 1988; Pierce 1990).

Genetic diseases have a major impact not only on the affected individual but on the parents, grandparents, siblings and children of the individual. The psychological impact of genetic disease varies with its severity and ability to treat, and within families there is the potential for individuals to react uniquely to corresponding situations. The suggestion of genetic disease can be perceived as a threat and may arouse emotions. During pregnancy such a threat may be particularly intense. Thus, "exploring these feelings (about the birth of a child with a birth defect) may be far more important than providing a statistical estimate of the risk, and somewhere during the counselling process there should be an opportunity to do so" (Fraser 1974 p 639).

4.2.1 Genetic counselling

The aims of genetic counselling have been outlined (Antley 1976; Fraser 1974). These are to help the individual: 1) comprehend the medical facts, including the diagnosis, probable course of the disorder, and available management; 2) appreciate the way heredity contributes to the disorder, and the risk of occurrence or recurrence; 3) understand the alternatives for dealing with the risk of occurrence or recurrence; 4) choose the alternative which seems appropriate to them in view of their risk and their family goals, and act in accordance with that decision; 5) make a healthy adjustment to their decision. Antley acknowledges that this definition is broad and encompasses long-term medical, psychological and sociological adaptations.

Nowadays, those involved in counselling individuals, at risk of developing or passing on a genetic disorder to their children, are aware that "it is no longer sufficient to be conversant merely with genetic and medical aspects of a problem. It is also important to be fully aware and appreciative of the psychological effects on the individual" (Emery 1984 p 5). For the midwife concerned with counselling for genetic disorders this reaffirms the value of the professions' commitment to the philosophy of holistic care.

The psychological aspects and techniques to manage individuals and couples within the field of genetic counselling have been presented (Emery and Pullen 1984). These are of particular relevance to this study given that CF is a genetic disorder. Moreover, studies in relation to other prenatal screening programmes, however relevant, do not involve screening for a single gene disorder, nor generating risks of occurrence as high as 1 in 4, nor do they introduce implications for other family members.

Two central themes in relation to genetic counselling are decision making and loss. Decision making can correspondingly be considered a central theme of prenatal CF carrier screening. Firstly, a woman must decide whether or not to be screened. If she is screened and receives a positive result she and her partner must decide if he, the male partner should be screened. If he is a CF carrier they must decide on the best available course of action. If a couple favour prenatal diagnosis and the fetus is shown to be affected, they are faced with a further decision: to continue or to terminate the pregnancy. Such a couple face profound loss. Loss has wide connotations in relation to genetic disease; loss of their child's health, loss of reproductive choice, loss of dreams and aspirations, altered body image and loss of self-esteem. For some young couples the birth of, or the risk of giving birth to a child with a genetic disease may be the first significant disappointment in their lives (Targum 1981).

Falek describes the sum and substance of genetic counselling as "the counsellor's ability to transmit genetic information about an inherited disorder of concern to the counsellee(s) so that it will be incorporated into decision making." (Falek 1984 p23). Falek recognises the difficulty of new information being received and incorporated on both cognitive and emotional levels at a time when individuals are under stress.

4.2.2 Loss in relation to genetic disease

The coping responses of individuals under stress in relation to genetic disease are described and compared to that of any loss or bereavement (Falek (1984). The initial response is shock and disbelief and frequently denial. Denial is used as a protective mechanism to maintain psychological equilibrium. The duration of the denial period depends upon the individual and the circumstances (Falek 1984). There is evidence that genetic information may be given too early during the coping process to be retained. One study reported that of 130 families followed up after having received genetic counselling for a serious genetic disorder diagnosed in their child, 25 per cent considered that counselling was given too early on in the grieving process to retain the information (Fraser and Levy 1972). Other researchers have found from 8 per cent to 15 per cent of families denied even having been counselled (Leonard et al 1972; Antley and Hartlage 1973).

Reynolds and co-workers carried out a retrospective study on 101 families who had received genetic counselling to assess the impact and effectiveness of the service. Sixteen per cent had either distorted or rejected the information and two per cent had repressed the entire counselling experience (Reynolds 1974). Several participants felt that a period of 3 to 6 months was needed to allow couples to adjust to the discovery of a birth defect

or genetic disorder and that for the duration assimilating genetic information or planning for the future was difficult.

Most couples do not consider the possibility that they may carry abnormal genes and often perceive their children as "self-enhancing extensions of themselves." (Antley 1976 page 112). They do not embark upon a pregnancy with the idea of taking a risk of having a child with a serious genetic disorder or congenital abnormality. When this occurs a couple's concept of an ideal family is shattered and major emotional adjustment is necessary. These findings raise concern in relation to prenatal CF carrier screening because couples who are identified as carriers are likely to be shocked and therefore may experience difficulty in absorbing information on which to base a decision about the future of their pregnancy. Moreover little time is available to make an emotional adjustment before reaching a decision.

The second stage of the coping process occurs when the individual begins to realise the circumstances of his or her stress and undertakes a cerebral approach to dealing with the situation. The individual responds by manifesting symptoms of anxiety including nervousness, over activity, irritability, headache, fatigue, insomnia, loss of appetite and somatic complaints (Falek 1984; Wilkinson 1992). Caplan alerts us to the fact that although anxious, the individual's need to formulate events initiates a search for information (Caplan 1964). Now the content of genetic information and discussion on available options and courses of action can be initiated. In so doing, the counsellor should be aware that those who are particularly anxious may continue to block information (Falek 1984).

The third stage of coping is manifest as anger. Anger may be directed outwardly or inwardly and in both cases there may be outward signs of hostility (Falek 1984;

Wilkinson 1992). Anger directed inwardly may result in guilt. Feelings of guilt in relation to genetic disease have been reviewed (Kessler et al 1984). These merit examination in relation to CF carrier screening and the care of those who receive a positive test result. Kessler and colleagues advise that general reassurance concerning feelings of guilt being normal is not sufficient; that time should be taken to establish exactly how an individual is feeling and to constructively tackle this in order to dispel these feelings. They submit that guilt is often confused with shame. Shame is defined as that which results from failure to reach goals whereas guilt arises when the individual feels that their behaviour has fallen short of their internalised ideal of what their behaviour should be. Therefore, they maintain that much of what is labelled guilt may be shame.

Distinguishing between guilt and shame is significant with regard to stress intervention. Parents normally invest of themselves (narcissistic investment) in a pregnancy (Raphael-Leff 1991). If the outcome is successful it leads to joy and fulfilment and parents feel pride in themselves and child. If pregnancy leads to disappointment and despair, parents feel a sense of failure and shame. Couples who receive a positive CF prenatal screening test result find themselves singled out and advised that they require special attention and counselling around their present and future reproductive decision, a decision over which most others normally exert autonomous control. It is possible that such couples could experience guilt and shame. Any measure of the psychological impact of prenatal CF carrier screening would require to take these feelings into account.

Kessler suggests a number of guilt alleviating tactics which the health care professional may use. Use of authority, where the professional uses the full force of his or her role to tell the couple they are guiltless. Normalisation, when the couple are reassured that their feelings are normal. Reframing can be used when a couple voice feelings that they "are responsible" by saying, "yes you are responsible - responsible human

beings," and reassures in this way. Limiting liability, simply acknowledges that the couple are responsible for bearing a child, but are not responsible for the genes they transmit to the child. Any or all of these strategies may assist in dispelling an emotion which can have a pervasive influence on a couple's perception of genetic information.

The fourth stage in the coping process, described by Falek, is the shift from guilt or anger to depression. This is the signal that the individual is entering the next stage in the coping process and is considered a "normal phase" in achieving restitution. Depression results from "the repeated frustration of attempts to resolve the problem." (Falek 1984 p 29). The individual will appear sad, withdrawn and seem uninterested in his or her environment and daily activities.

The final or fifth stage of the coping process as described by Falek is one of psychological homeostasis. Now the individual is likely to be receptive to the genetic counselling process. Falek cautions that reversion to earlier coping phases during this stage is a normal part of attaining psychological homeostasis.

The conceptual model outlines those factors which determine an individual's response to a stressful event (Cochrane 1983). The contribution of Falek's work to this study is to describe in more detail those manifestations of stress which can be expected and which need to be recognised and acknowledged before intervention strategies can be initiated to assist coping.

4.2.3 Decision making in genetic counselling.

The objectives of genetic counselling may be clear but what is not so apparent is how couples come to make their choice or decision. Perceived risk rather than actual risk of occurrence is thought to assume more importance in the decision making

process (Marteau and Slack 1992a). It is believed that couples do not base their decisions solely on medical facts, but on complex, deeply personal interpretations of these facts (Wertz et al 1984). Anecdotal evidence that instinct and emotion rule, rather than risk factors and percentages, was given by a young doctor who described how she and her partner, faced with a positive maternal serum alpha-fetoprotein screening result, were overwhelmed with the emotional considerations of potential fetal abnormality and unable to base their decision to undergo prenatal diagnosis on the factual data offered (Anonymous 1989).

The conclusions drawn about the impact of genetic disease are: that couples who learn that there is a high risk of their child being born with a genetic disorder have to come to terms with a sense of loss and have to consider the options open to them; they experience shock and frequently guilt and shame. Counselling can help couples firstly come to terms with their emotions, to understand the disorder in question, the options open to them and the implications of these options. Couples who choose to continue a pregnancy without intervention need help to prepare for their possible future role as parents of a child with special needs and will require further consultation and counselling. Couples who choose fetal diagnostic testing will need psychological counselling during the diagnostic process. Those who receive reassurance regarding the health of the fetus will require further consultation and counselling to allay any residual concerns. Those confronted with a positive test result may require crisis counselling and help in reaching a decision regarding the continuance or termination of their pregnancy. Regardless of their decision ongoing support and counselling would need to be made available in the months ahead.

4.3 Prenatal screening and diagnosis

One of the fastest growing areas of medicine has been the development of techniques for fetal diagnosis of genetic diseases and congenital malformations, yet relevant social science research has been limited to small and often selective samples (Richards 1987). The early studies to which Richards referred were, nevertheless, testimony to the psychological impact on women who experienced these new prenatal tests (Beeson and Golbus 1979; Farrant 1985; Rothman 1988). Since then, an increasing amount of research into the psycho-social aspects of prenatal screening and diagnosis have confirmed and augmented their findings.

4.3.1 Factors influencing uptake of prenatal screening and diagnosis

From some of these studies it is useful to focus on data pertaining to factors which appear to influence uptake of testing as these may also apply to prenatal CF carrier testing.

Availability

Firstly, a test may be available in theory but not in practice (Holland and Stewart 1990). The prenatal screening tests which women experience, to some extent, depend on the region in which they live and the hospital at which they book. UK health regions vary in the tests offered and what is available in one hospital may not be available in another (Cuckle et al 1989). Consultant obstetricians differ in their stance with respect to timing and frequency of ultrasound scanning and the maternal age at which amniocentesis is suggested. A consultant's attitude toward granting a request for prenatal screening and diagnosis can vary, especially towards the 'anxious woman' who may request a test on the grounds of wishing reassurance, rather than because she is at increased risk of fetal abnormality (Sjogren and Uddenberg 1990).

Availability of a test may be dependent on the gestation of a woman's pregnancy. For example, if a woman presents too late in pregnancy maternal serum alpha-fetoprotein screening (MSAFP) (most accurate between 16 and 20 weeks) would not be available. Although accuracy of the CF carrier test would not be influenced by pregnancy gestation, selective abortion would. There is substantial evidence that therapeutic termination for fetal abnormality in the second trimester has a profound and lasting effect on women (Blumberg et al 1975; Donnai et al 1981; Adler and Kushnick 1982; Leschot et al 1982; Jorgenson et al 1985; Lloyd and Lawrence 1985). Men too have been shown to experience symptoms of stress subsequent to their female partner having undergone termination of pregnancy for fetal abnormality (White-Van Mourik et al 1992). Consequently, women presenting after 18 weeks of pregnancy were advised against CF carrier testing, resulting in around 10 per cent of women not being screened for this reason (figure 2.5. page 46).

Attitude and knowledge of health care professionals

The attitude of obstetric staff toward a prenatal screening test can influence uptake. For example, a test may be offered as if it is routine, rather than voluntary and requiring a decision. A study recording the offer of maternal serum alpha-fetoprotein screening by midwives and obstetricians found that in half of 102 consultations the test was presented as a routine test with little explanation (Marteau et al 1992c). Limited professional awareness, and severely limited technical, educational and counselling resources are also blamed for deficiencies in prenatal screening and diagnosis. Shortcomings can be blamed to some extent on under funding but missed opportunities arise because basic clinical genetic knowledge is lacked by both the medical and nursing profession (Guilbert and Cheater 1990; Johnston 1990; Royal College Physicians 1989).

Farrant (1985) observed that obstetricians perceived the significance of prenatal screening as one of diagnosis and abortion of affected fetuses. In contrast, most women perceived it as a means of receiving reassurance. Among obstetricians there are those who hold the view that "prenatal diagnostic procedures may be of significant benefit to both mother and child even when the detection of an abnormality would not lead to pregnancy termination" (Clark and DeVore 1989 p 1035). However, there are those who are reluctant to carry out a risk-associated procedure under these circumstances and will attempt to discourage a couple opposed to termination of pregnancy from pursuing prenatal diagnosis (Crawfurd 1983; Thorp and Bowes 1989). Similarly the attitude of the midwife regarding which conditions she regards as serious and considers justify termination of pregnancy may influence the way she presents a particular test (Marteau and Slack 1992a).

Attitude and knowledge of women

Surveys have shown that a majority of women wish prenatal screening. Attitudes toward maternal serum alpha-fetoprotein screening in a cohort of 2254 pregnant women found 98 per cent wished to be screened (Bennett et al 1980). In a later survey 98 per cent of 1235 women booking for prenatal care were in favour of MSAFP screening (Kyle et al 1988).

Whether a woman accepts MSAFP screening or amniocentesis is believed to depend upon her knowledge of the test, her perception of the risk and burden of having an affected child, the reliability of the test and in the case of amniocentesis her concern about miscarriage (Marteau and Slack 1992a). Understanding the risk of fetal abnormality was cited as an important factor in a woman's decision to undergo prenatal screening or fetal diagnostic testing (Marteau et al 1988a), but more recent research by Marteau indicates that a woman's perception of her risk rather than knowledge of actual

risk, influences her decision regarding the uptake of amniocentesis for reasons of advanced maternal age (Marteau et al 1991a). Attitude to termination of pregnancy is a major influencing factor as to whether a woman declines or accepts a prenatal screening test (Davies and Doran 1981; Faden et al 1987; Kyle et al 1988).

Studies of parental perceptions of genetic disease have discovered that burden of the condition influences whether women accepted or declined amniocentesis for diagnosis of a serious congenital abnormality. A group of 252 women, who had received genetic counselling after the birth of a child with Down's syndrome or genetic disease amenable to prenatal diagnosis, were interviewed to explore factors which influenced the decision of 202 to accept and 50 to decline amniocentesis in a subsequent pregnancy (Ekwo et al 1987). Those conditions which resulted in a prolonged illness or early death were considered to be most serious and warranted prenatal diagnosis, those resulting in a physical handicap or a facial disorder least burdensome, while those conditions associated with childhood mental retardation fell in between. Thus women who perceived a disorder as too burdensome accepted the offer of prenatal diagnosis. The researchers concluded that couples' perceptions of how they would cope with the medical and social consequences of a disease should be an integral part of counselling. This study corresponds to that of the findings of Decruyenaere and co-workers (1992) which was discussed on page 73 of this text. and in which the perceived burden of CF was associated with choosing the option to forego pregnancy in the event of confronting a 1 in 4 risk of a child with CF.

Women will vary in their perception of which conditions justify termination of pregnancy. Faden and co-workers (1987) questioned 490 women, among whom 300 accepted the offer of MSAFP screening, about their attitude on abortion for varying degrees of disability and certainty of diagnosis. A woman's religious beliefs were found to

influence attitude. The more severe the disability the more termination of pregnancy was considered justifiable. Notably certainty of diagnosis was found to be an important factor in a woman's decision to terminate a pregnancy. For example, 6 per cent of respondents considered termination of pregnancy justifiable if the diagnosis of a neural tube defect was 25 per cent certain, this figure rose to 56 per cent if the diagnosis was 95 per cent certain and to 80 per cent if certainty of diagnosis was 100 per cent.

It is thought that women frequently participate in prenatal screening programmes in order to avoid regret at not having done so (Tymstra 1989) and because the availability of screening raises their awareness of fetal abnormality (Rothman 1988). Many women will enter a prenatal screening programme with the primary goal of being reassured about the normality of their fetus (Davies and Doran 1981). In a majority of cases the experience results in reassurance but for the 5 to 10 per cent who receive bad news it causes distress. It is, therefore, critical that women are aware of the reason for being screened, the chances of receiving a positive test result and are encouraged to give some thought as to what they might do if faced with a poor result (Donnai et al 1981).

4.3.2 The psychological impact of prenatal screening.

The similarity between MSAFP screening and prenatal CF carrier screening is that all pregnant women are potentially eligible for screening. The major differences are that the CF test can be performed outwith or during pregnancy, whereas MSAFP screening cannot be carried out before 16 weeks gestation of pregnancy. In addition the significance of a positive test result differs: a positive MSAFP result is a fetal indicator of possible neural tube defect or Down's syndrome; whereas the CF test result screens the mother not the fetus. Nonetheless, a positive CF test result could be perceived as a threat

to the fetus and generate a similar emotional response to that shown by women who have received a positive MSAFP result.

Whether offering prenatal testing is, in itself, stress provoking has not been fully investigated (Marteau and Slack 1992a). One study measured levels of anxiety in women who accepted MSAFP screening compared to women who declined the test, and found no significant difference (Berne-Fromell and Kessler 1984). Another study by Burton et al (1985b) also compared similar groups of women and suggested that those who were screened tended to be less anxious than those who declined. One reason for this may be that women who feel anxious toward screening may avoid prenatal tests on the grounds that it could generate stress. However, because of the lack of research it is difficult to draw firm conclusions.

A significant finding in relation to prenatal screening is: that women are more distressed if shown to be at risk of fetal abnormality through a screening programme, compared to those who were previously aware of their risk (Farrant 1985; Tsoi et al 1987b). The shock element is thought to be critical. Women who know that there is a risk of a fetal disorder prior to conception may feel differently about the fetus than a mother who learns of this possibility during pregnancy (Richards 1987). An additional distinguishing factor is that women who know they are at risk are more likely to be knowledgeable about the condition and are not confronted with having to assimilate information when they are distressed (Mouzouros et al 1980).

Studies devoted to assessing the psychological effects of prenatal screening and fetal diagnosis have been reviewed (Green 1990b; Tunis and Golbus 1991). Level of anxiety has been the variable most frequently measured and The State-Trait Anxiety Inventory (STAI) (Spielberger et al 1983) has been the measure of choice in a majority of studies.

Anxiety has been shown to rise when a positive test result is received, and subside after a negative outcome is known (Fava et al 1982; Verjaal et al 1982; Robinson et al 1984; Burton et al 1985a; Tsoi et al 1987b). In one study reassessment of anxiety later in pregnancy showed a further rise (Tabor and Johnsson 1987). Variations within studies may reflect the way in which women are managed, particularly with regard to how results are conveyed and the quality of counselling at that time. Summarising genetic information in writing for patients has been recommended (Hecht and Holmes 1972; Reynolds et al 1974).

There are numerous studies which attest to the distress which a positive prenatal screening test result engenders. MSAFP screening began in the late 1970's with little or no attention being paid to the psychological impact of testing, rather concentrating on availability and uptake of screening. The first study devoted to women's experiences of the MSAFP screening process was by Fearn and colleagues (1982). The results showed high levels of anxiety in those women who received a positive test result. Anxiety levels remained high among those who experienced a false-positive result and were subsequently reassured. Results such as this led to the concern that having once cast doubt on the health of the fetus it may be difficult to completely reassure a woman.

A British study assessed the knowledge, attitude and levels of anxiety among 161 women aged less than 38 years who were offered MSAFP screening. Twenty per cent declined the offer and differed from those who accepted in their attitude to termination of pregnancy. They were also less likely to attend antenatal classes although no difference was detected in the health-related behaviour of the two groups. Levels of anxiety were monitored by the Spielberger State-Trait Anxiety Inventory (STAI) (Spielberger 1983) which was administered at booking, 17 weeks, 28 weeks, 38 weeks gestation and 2 days

postpartum. Of the 128 women who were screened, 10 received a positive test result and after retesting they received a negative test result. All 10 women manifested anxiety at the time of receiving their positive test result. Subsequently they showed a decline in STAI scores at the remaining three assessment points similar to women who were only tested once. Conversely, those who declined the invitation to be screened showed a rise in anxiety at 38 weeks gestation and were somewhat less positive in their attitude toward the pregnancy (Marteau et al 1989b).

Studies from other countries have reported similar findings: a rise in anxiety at the time of receiving a positive test result but a return to control levels once a negative result is received; and a tendency for screened women to be less anxious during the later stages of pregnancy compared with those who were not screened (Berne-Fromell et al 1983a, 1983b, and 1984; Burton et al 1985a and 1985b)

The implications of MSAFP screening have widened since low levels of MSAFP were found to be associated with Down's syndrome. Results from one study compared women who were identified at increased risk of Down syndrome through MSAFP screening and women over 35 years. Findings revealed the screened group to be significantly more anxious than those who had not undergone screening despite their having similar statistical risks (Abuelo et al 1991). Again the element of shock was thought to be a major factor, supporting earlier findings. (Farrant 1985; Tsoi et al 1987b).

Women receiving false positive MSAFP results indicative of Down syndrome were compared with a group aged 38 years and older. The younger screened group were significantly more anxious both at the time of receiving their test result and three weeks later when a normal result was known. Older women who had been routinely

treated as being at risk of having a baby with Down's syndrome were considered to be better informed and prepared for bad news (Marteau et al 1988b). The conclusion drawn was that understanding of screening tests is a major factor in preventing an adverse psychological response to a positive screening test result. Those previously aware of their increased risk were better able to formulate information and prepare psychologically for prenatal diagnosis than those younger women who, detected through a screening programme, were taken unawares and had to assimilate information and make a decision whilst under stress.

It seems probable that most women will receive news that they are a CF carrier with shocked surprise. If they feel threatened, or perceive that their pregnancy is threatened they are liable to manifest symptoms of anxiety, and if they suffer from anticipatory loss then they may manifest symptoms of depression. If their partner receives a negative test result the evidence is that their distress will subside. However, it may resurface because the risk of a CF child cannot be eliminated only reduced. To what extent couples in this situation will dwell on the residual risk, which has after all increased from their starting risk is difficult to anticipate. There is certainly evidence that if a woman perceives that doubt has been cast on the health of the fetus then it is difficult for her to be fully reassured. CF carrier screening differs from other prenatal screening tests in that it does not screen the fetus only the parents, but this may present a subtlety which some could find difficult to understand. Another difference is that these couples where one is a carrier and the other screens negative are left in limbo because there are no further tests which could be offered to further clarify the position. This contrasts with maternal serum screening tests for Spina Bifida and Down's syndrome where a positive screening test result can be clarified by offering ultrasound examination, amniocentesis or chorionic villus biopsy to establish the status of the fetus (Crespigny and Dredge 1991).

4.3.3 The psychological impact of prenatal diagnosis

Early studies on the emotional effects and experiences of women who had undergone amniocentesis were retrospective and tended to be based on anecdotal evidence. Without exception they showed that women found the procedure and the period awaiting the test result stressful (Robinson et al 1975; Chevrin et al 1977; Goodmillow et al 1978; Vinson et al 1980; Farrant 1980). Correspondingly, studies measuring the psychological impact of prenatal diagnosis have focused on levels of anxiety generated by the diagnostic procedure (Astbury and Walters 1979; Beeson and Golbus 1979; Fearn et al 1982; Black and Furlong 1984; Burton et al 1985 a,b; Phipps and Zinn 1986; Tabor and Jonsson 1987; Marteau et al 1989b; Abuelo et al 1991). Of 90 women who underwent amniocentesis because of a raised MSAFP result, 68 per cent felt their health had suffered during that period. This compared to 22 per cent of women who underwent the same procedure for reasons of advanced maternal age (Farrant 1980), a finding which has resulted in the previously mentioned theory that women who already perceive themselves to be at risk of fetal abnormality find prenatal diagnosis less stressful than those ascertained through a prenatal screening programme.

The emotional response of a group of women undergoing amniocentesis for advanced maternal age and a control group drawn from non-participants were studied (Phipps and Zinn 1986). Amniocentesis subjects were more anxious at the time of the procedure than control subjects, but became less anxious than their control subjects at the time of receiving a negative amniocentesis result. Notably, both subjects and controls with a history of previous fetal loss from spontaneous abortion showed the highest anxiety scores.

A follow up study of 164 couples to evaluate reproductive decision making after genetic counselling, found that 43 per cent had experienced difficulty making a decision. Among those 109 (66%) who had decided to have further children 45 per cent of couples who knew that prenatal diagnosis was available had found the decision difficult, compared to 23 per cent of couples for whom no prenatal diagnostic test was available (Frets et al 1991). The issues facing couples for whom prenatal diagnosis is an option is discussed by Rothman (1988) in an anthology of accounts of women who experienced prenatal diagnosis. The major issue is that of abortion which is assumed to be an integral part of prenatal diagnosis. Rothman postulates that the public are more accepting of abortion in this circumstance and in turn presume it to be less traumatic psychologically. She argues that in no sense does it make abortion any easier as women with an unwanted pregnancy see it as an accident "a by product of contraception that did not work" (p5) and the abortion as a solution, whereas women who plan a pregnancy want and perceive the fetus as their baby and to abort their baby even if it is imperfect is a tremendous struggle. She quotes one woman who had undergone amniocentesis:

"By the time the results came in the baby had been leaping in my womb for a month.....During one of the sleepless nights before the results were in I decided I would raise the child if it looked like E.T." (p7).

The period of waiting for the test result and the burden of selective abortion cannot be underestimated. Rothman's findings contradict any idea that prenatal diagnosis may offer an easy way out.

The conclusions which can be drawn from reviewing the impact of prenatal screening and diagnosis are foremost, that whether offering a prenatal test in itself creates stress is difficult to answer because studies have not directly addressed this question. For the present, what little evidence there is would suggest not. It would appear that women

who receive a positive test result are more likely to experience a stressful reaction if they are found to be at risk of fetal abnormality through a screening programme and did not previously consider themselves to be at risk. Perception of risk is, therefore, an important factor and allied to this younger women are likely to perceive themselves at lower risk than are older woman. The evidence that women who have suffered a previous fetal loss find prenatal diagnosis particularly stressful is a notable finding and it seems reasonable to theorise that this group may also find prenatal screening stressful.

The impact of prenatal screening and diagnosis upon a woman or couple seems to be influenced by a number of factors: their reason for undergoing the test; their understanding of the test result; and the care they receive from health professionals (Marteau 1991b). Some women will receive written information and counselling before deciding to undergo these procedures and afterwards, while others will not. Lack of understanding about the test is liable to contribute to a stressful reaction.

4.4 Early infant loss

Early infant loss includes all reproductive losses: spontaneous abortion; death of an infant; termination of pregnancy; the birth of a child with a congenital or genetic abnormality which constitutes loss of infant health or normality; and loss through relinquishing a baby for adoption. Loss associated with the latter often fails to be recognised. The assumption that voluntarily parting precludes the need to grieve (Mander 1991a).

4.4.1 The psychological impact of early infant loss

It is now recognised that the parent who loses a baby early on through spontaneous or therapeutic abortion grieves as those who lose an infant at or after birth and should

be offered the same opportunities afforded to couples losing a baby in later pregnancy or post-delivery (Bluglass 1984; Iles 1989). Iles found evidence that psychiatric morbidity was higher among those where termination was carried out later in the second trimester. Moreover, levels of anxiety and depression were greater if the fetal condition was compatible with extrauterine life; for example when a baby had spina bifida rather than anencephaly (Iles 1989). This finding has particular relevance to prenatal carrier screening for CF which offers the potential to prevent the birth of a child suffering from a condition where, increasingly, affected individuals survive into adulthood. It demonstrates the need for stress intervention strategies by midwives in caring for those who undergo termination of pregnancy for conditions such as CF. There is evidence that midwives experience difficulty in initiating stress interventions which might help the bereaved mother adjust to her loss. (Mander 1991b). Midwives feel an underlying awkwardness which stems from having to adjust from caring for the mother who has experienced a successful pregnancy to caring for the bereaved mother. Allied to this is a reluctance to burden the grieving mother with decisions about her care (Mander 1991b).

A programme of prenatal screening or prenatal diagnosis should have, as part of its infrastructure, a termination service. This service should ensure that where a fetal disorder exists the most appropriate technique for terminating the pregnancy can be offered. The primary objectives of the service and various procedures available to terminate a pregnancy are outlined by Mackenzie (1992). The primary aims are: that therapeutic termination can be carried out without contravening the law; with minimal immediate and long-term risks to the mother's health; in a manner which is acceptable to the mother; and that prenatal diagnosis can be confirmed following the termination. The psychological consequences of termination for fetal abnormality indicate that patients frequently experience residual feelings of guilt and distress relating to failure to produce a healthy child and the decision to end the pregnancy (Iles 1989).

These feelings are more acute after therapeutic termination than those carried out for social reasons (Blumberg et al 1975; Donnai et al 1981; Lloyd and Laurence 1985; Iles 1989). Reasons for this were recounted on page 92 as exposed by Rothman (1988).

A recent retrospective study on a cohort of women who underwent therapeutic termination of pregnancy in the second trimester merits review (White-Van Mourik 1992). Twenty per cent of 84 women interviewed claimed to experience tearful episodes, sadness and irritability 2 years after their loss. Male partners reported increased listlessness, loss of concentration and irritability up to 12 months after the termination. Twelve per cent of couples reported marital disharmony. Younger women (less than 20 years) or immature women were more vulnerable to prolonged grieving. Notably, younger women also reported feeling they had been pushed into the decision by well meaning individuals around them. Couples identified several themes as important. They wished their feelings of grief acknowledged and time needed to come to terms with their loss. They wanted information and communication about the fetal disorder, implications for future pregnancies, prenatal diagnostic and termination procedures, and information regarding post-termination psychological and social sequelae. They expressed need for reassurance that psychological symptoms were a normal aftermath of termination, and sought advice on coping strategies and hope regarding future pregnancies.

4.4.2 Nursing intervention for those who suffer early infant loss

Although bereavement counselling has similarities to genetic counselling it involves more listening and less talking. The individual or couple are given time to reflect upon the circumstances of their loss. Parkes (1975) describes grief as a process, not a state, which can be expressed in different ways at different stages in the process.

Moreover, the loss experienced by bereavement frequently results in secondary loss such as dissolution of reproductive choice, dreams and aspirations.

The role of the nurse in counselling and supporting couples who have experienced early infant loss has been sensitively outlined (Hopper 1991). For those couples who face therapeutic termination of pregnancy it is a sad and lonely episode with an inevitable unhappy end. Hopper describes the basic needs of most couples as: trust in those caring for them; to reflect upon their loss; to be allowed to remember; and to assimilate the events. The importance of a couple's unique circumstances which in turn determines their own special needs are emphasised and attention drawn to the fact that a couple is composed of two individuals whose reactions and needs may vary. Hopper advises that both partners should be consulted individually to determine what they feel their needs are at each stage of the episode.

Hopper also highlights the importance of encouraging couples to reflect upon their loss and to express what the loss of their baby means to them individually and as a couple. The midwife can, she suggests, best achieve this by giving couples a licence to talk about the hopes and aspirations they held for their child. The creating of memories is considered beneficial to the grieving process and encouraging a couple to accept tangible evidence of their experience such as a sonogram picture can be consoling. Finally assimilation of the events is a normal and necessary process in coming to terms with grief. As Hopper states, "every story needs a beginning, a middle and an end." (page 21). This means allowing a couple the opportunity to recount the sequence of events in the days, weeks and months after termination so that they may make an adjustment to their loss.

4.5 Pregnancy

The CF carrier test is offered to pregnant women at their first antenatal clinic visit (average 12 weeks gestation) when the early psychological and physical changes associated with pregnancy are experienced. The literature about pregnancy is immense and this literature search concentrated particularly on the early stages of pregnancy and on the stresses which the state of pregnancy itself could create.

Pregnancy is seen as a life event and transitional period when a woman prepares for the maternal role (Blumberg 1984). This transition involves widespread psychological changes and a reappraisal and redefinition of identity, particularly in a first pregnancy when there may be a certain amount of conflict and stress. Physical discomfort is reported by substantial numbers of women. In a study of 105 pregnant women 68 per cent complained of lethargy, 68 per cent of leg cramps, 66 per cent of urgency of micturition, 48 per cent of backache, 46 per cent of breathlessness and 43 per cent of indigestion. Apart from these physical discomforts, the same subjects being interviewed on a number of occasions during pregnancy also reported crying (53%), misery (50%), nervousness (34%) and worrying (29%) as psychological manifestations of the second and early third trimester (Wolfkind and Zajicek 1981). Green found that early in pregnancy 74 per cent of women reported feeling happy but 46 per cent felt anxious. Their major worries were miscarriage (31%), something being wrong with the baby (28%), financial concerns (26%), concerns about going into hospital (14%) and internal examination (18%) (Green 1990d).

According to a recent study involving obstetricians, midwives and pregnant women, attitudes between the three groups differed with regard to the riskiness of pregnancy. Obstetricians perceived pregnancy as significantly more risky than did midwives and pregnant women. Female obstetricians perceived pregnancy as

significantly more normal than their male colleagues. In addition, the views of obstetricians and midwives tended to be associated with number of years practising, with longer practising obstetricians seeing pregnancy as 'more risky' and longer practising midwives perceiving it as 'less risky' (Schuman and Marteau 1993). Offering prenatal screening for fetal disorders emphasises afresh the risks associated with pregnancy. For the midwife being asked to present yet another prenatal screening test, in the form of CF carrier screening, means that an increasing amount of the booking procedure is spent discussing abnormality rather than normality.

4.5.1 The Psychological processes of pregnancy

Each woman's experience of pregnancy takes place within the context of different emotional, psycho social and physical circumstances. Pregnancy means that the woman, especially the primigravida, has to " find a way of incorporating the idea, and bearing the reality of another being sharing her inner space and becoming part of her internal world" (Raphael-Leff 1991 page 45). In traditional societies there are ceremonies and rituals which help a woman to adjust to these experiences, but in our society each pregnant woman has to find her own means of coping (Jiminez and Newton 1979). Field (1990) states that the whole area of psycho social support has been neglected in the provision of antenatal care and states that "midwives need to understand the processes involved in becoming a mother. " (p 219). Field argues for continuity of care which will allow a woman to build a trusting relationship with the same midwife and discuss concerns relating to all stages of pregnancy. In turn this will allow midwives to make a significant contribution to the psychological care of mothers. Sweet (1988) advises that by giving the mother an opportunity to address and understand her own fears and concerns, the midwife can help a woman to increase her control. Feeling in control have been shown to be associated with positive psychological outcomes of childbirth (Green 1990a).

During the early months of pregnancy unfamiliar physical changes can be especially disturbing and physical examinations may be seen as invasive and particularly stressful to the primigravida (Green 1990d). Emotional disequilibrium causes inexplicable mood swings, intense urges, memory lapses and sudden flashes of insight. Frequent, vivid dreams may be disturbing to some women and in waking life she may relate more keenly to her environment (Lederman 1984). Some women never experience negative emotions during their pregnancy but rather a heightened feeling of psychological well-being (Elliot et al 1983; Condon 1987).

The diversity of the psycho social processes of pregnancy are outlined by Raphael-Leff (1991) and using anecdotal evidence she highlights how each woman responds in her own individual fashion. This is an important consideration for the presentation of prenatal screening as each woman will view it from her own particular perspective. This could in turn determine her overall response in terms of uptake of the test, her expectations of the test and the psychological impact of the test. Raphael-Leff also focuses on defining women who are particularly at risk of a stressful reaction (Raphael-Leff 1991) and who, she suggests, can be identified by careful history-taking at the antenatal booking clinic by sensitive questioning, and observation by midwives during subsequent antenatal encounters. Careful assessment of a woman's support systems, her feelings toward pregnancy and changes in lifestyle is considered paramount in providing supportive care and assisting women to make the transition to successful motherhood (Field 1990). Women who enter pregnancy already experiencing stress or for whom pregnancy induces stress will also perceive the advantages and disadvantages of screening from their own particular perspective. The cause of their stress or their reaction to it may influence their attitude. Indeed being aware of any stress, the amount of it and nature of it could influence pre-screening counselling. If a woman were identified as a CF carrier, previous awareness of ongoing stress could be

taken into account in assessing her emotional response and in her ongoing management during the screening process.

Jones (1990) proposes that psychological self-assessment should be an integral part of antenatal care. A descriptive study was carried out by Jones to assess if women would benefit from completing a self assessment form designed to identify their concerns about pregnancy and to identify the type of woman best suited to self-assessment. Twenty five women who attended an antenatal clinic, who were admitted to an antenatal ward, or were visited during the postnatal period within their own home were asked to complete a self-report assessment form over a period of a week. The form consisted of a series of headings which were explained to the women by the researcher. These were: self concept, body image, sexuality affect and mood, pain, spiritual feelings, ability to cope, stress reactions, role function and relationship with others. Participants were asked to complete details about their educational background and career to " identify the type of woman most suited to self-assessment." (page 37). Other than reporting that these data did not influence the number who responded to the study, Jones fails to report whether any correlation was observed between educational background and career and women's responses to the discrete areas of psychological assessment. Moreover, there was no indication that data such as age, parity, marital status or obstetric history had been collected which might have influenced the response of women to the assessment schedule. For example social class was found to influence the attitude of a group of working-class women in Glasgow who considered childbirth "a hurdle to be surmounted on the way to motherhood" (McIntosh 1989 page 193). How a woman responds to prenatal screening for CF may be influenced by her psychological attitude to pregnancy and this in turn may be influenced by her educational and social background and previous obstetric experience. Situations which could cause women to present at the booking clinic suffering from psychological disturbance were, therefore, reviewed. The

researcher was guided by those categories of women cited by Raphael-Leff (1991) as being particularly at risk of a stressful reaction during pregnancy.

4.5.2 Stress in pregnancy

The untimely pregnancy

At present a quarter of all births in the UK are to single women and of these a quarter are to mothers under 20 years of age. Most are to teenagers over the age of 16 years (Lawson 1990). The emotional upheaval of pregnancy and adolescence together may precipitate a crisis (Bury 1984). Age has been cited as a factor in a woman's prenatal adjustment to the maternal role. Women below the age of 25 years were significantly more likely to report their pregnancy as unplanned, had significantly fewer years of education, relationships of shorter duration, and were less likely to have read about pregnancy and fetal development (Gottesman 1992). Younger women tended to rely more on the experiences of their mothers and sisters as guides for their own pregnancy experiences, whereas, older women more often relied on the experiences of friends. Younger women were less likely to notice patterns in their partner's response to the pregnancy and were less likely to describe having any relationship with their fetus, being more likely to describe their feelings as "scared and apprehensive."

Conversely, the older woman too may experience additional physiological and psychological stresses (Michelson and Gee 1984). This is often because they choose to delay having a baby in order to establish a career, or because they have not met the right partner, or cannot afford it, or because they do not feel ready for parenthood. She may feel torn between the conflicts of the biological clock and pursuing a career and worry about the increased risk of age-related fetal abnormality, of coping physically with pregnancy, and how she will cope with sleepless nights and a career. Their age and

experience of life makes them aware of the risks of pregnancy, moreover, they are more likely to experience greater medical intervention than younger women. Women report feeling that they were viewed negatively by the medical profession and society (Berryman and Windridge 1991) suggesting an element of perceived irresponsibility which apparently contrasts with actual findings which show that pregnancies of older primigravidae are carefully planned (Kitzinger 1982).

Unplanned pregnancies can pose moral and emotional dilemmas, economic and career sacrifices, and change in life style. An unsound relationship may not stand the strain and in what has appeared to be a happy marriage or partnership discord may arise. An ongoing pregnancy does not automatically mean acceptance and some women will deny the pregnancy exists and may appear quite late for antenatal care. Lester and Farrow (1988), studied women who had presented for antenatal care at 18 weeks gestation or later. Fifty two per cent claimed their pregnancy was unplanned. The proportion of unplanned pregnancies was greater among those belonging to the lower social class groups and among women whose partners were unemployed. Failure of contraception was not a problem, with 79 per cent of participants confessing to having themselves failed to use any form of contraception, or admitting to inconsistent use of contraception.

An untimely pregnancy may be too soon after a stillbirth or a neonatal death. If mourning is properly achieved a pregnancy can offer consolation and fulfilment, however, a new pregnancy can cut short the grieving process and predispose to psychological disturbance. Other family members such as grandparents may mourn deeply or push for another pregnancy (Oglethorpe 1989). Studies show that in a hasty subsequent pregnancy it can be difficult for a woman to come to terms with her feelings toward her lost infant, and adjust to her thoughts and feelings about the new baby with whose

safety she is concerned (Bourne and Lewis 1984). Good midwifery care in a subsequent pregnancy is reassuring and part of that care should allow couples to express specific anxieties and ask questions. Bourne and Lewis advise that persistently repeated questioning can indicate underlying anxieties or grievances which are being missed and need to be addressed by the midwife.

Wolff examined rates of conception following cot deaths and found 34 per cent of their sample of mothers were unable to conceive after trying for one year and 31 per cent had spontaneous abortions, compared with an expected rate of infertility of 10 per cent and a spontaneous abortion rate of 12 to15 per cent (Wolff et al 1970).

Pregnancies which occur as a result of rape or incest can generate feelings of hate or repulsion toward the fetus (McMahon 1992). Pregnancy and birth can also evoke memories among women who have suffered sexual abuse causing anxiety and depression, alongside emotional conflicts and worry about the baby. McMahon urges midwives to take on the issue of sexual abuse rather than ignoring it, and to be aware that therapy is available.

Overvalued pregnancies

Frequently these are long-awaited conceptions as a result of infertility, recurrent congenital abnormality or chronic disease, or a primigravida in whose immediate family a pregnancy or delivery has failed. Anxiety among these women is common (Oglethorpe 1989). In Britain it is believed that more than one in ten couples experience difficulty in either achieving or having a live born child (Page 1988). Studies indicate that couples who undergo infertility investigations and subsequently encounter prenatal diagnostic procedures experience elements of the psychological trauma associated with their infertility (Sandelowski et al 1991). Although, the older mother

may be anxious about the increased risk of obstetric problems, she may be ambivalent about undergoing invasive tests with an associated risk of miscarriage particularly if she experienced difficulty in achieving her pregnancy (Lilford 1991).

Women who are themselves adopted, though they yearn for a child of their own, often feel disadvantaged because they lack knowledge about their mother's or immediate family's obstetric history. Moreover, pregnancy may trigger emotional repercussions arising from her own adoption (Stewart 1992). Research into mothers who relinquish their baby for adoption reveals a reluctance by midwives to discuss interventions which might assist a woman cope with her grief response (Mander 1991b). Thus a mother may embark on a subsequent pregnancy not having come to terms with the loss of her baby.

Couples who already have a child with a chronic illness or disability may be particularly sensitive about prenatal screening and diagnosis. A study exploring attitudes toward alpha-fetoprotein testing among parents who already had a child with spina bifida, found that the majority would have accepted testing had it been available to them, and a large majority stated they would use testing if they became pregnant again. However, these couples expressed strong feelings in relation to whether they viewed abortion as an option. Half stated they would not consider terminating an affected pregnancy and many found themselves faced with anguish, confusion and ambivalence regarding termination (Van Cleve 1993). Whyte (1992) similarly found among couples who had a child with CF that the prospect of prenatal diagnosis in a subsequent pregnancy created painful dilemmas because it implied a devaluation of the worth of their affected child. These findings demonstrate a need for midwives to be aware of the sensitivity of information about prenatal testing among couples when there is an affected child in the

family. This need not necessarily mean a child belonging to the couple themselves, the affected individual may be a sibling or more distant relative.

Unsupported women

Woman who are alone in their pregnancy either because of death, divorce or abandonment by their partner are already undergoing a loss-related crisis and are particularly vulnerable. Conversely, there may be a husband or a partner but he may be unsupportive emotionally, absent, or worse abusive (Bewley and Gibbs 1991; Bohn 1990). Many, if not most, battered women are of childbearing age and some studies quote very high incidence figures. Campbell (1986) estimated that in the United States one in every 50 pregnant women may be beaten. Andrews and Brown in 1988 conducted research into marital violence in Islington, London. Of 286 working class women with children who were interviewed, 25 per cent had been subjected to violence by their partner. Pregnancy is reported to be a time when abuse begins or escalates (Hillard 1985). Attacks to the abdomen can cause miscarriage, placental abruption and premature labour or stillbirth. Frequently abused women exhibit stress and clinical depression and reluctance to discuss the issue (Andrews and Brown 1988).

Concurrent life events

Concurrent life events, such as moving house, or a new job, are major stressful events in anyone's life, but coupled with pregnancy they can prove too much and may lead to crisis (Robinson 1984). Pregnancy related events such as spontaneous abortion, stillbirth, fetal abnormality, or positive screening test results invoke a grief reaction which needs to be worked through (Hopper 1991).

Substance abuse and risky behaviour.

Eating disorders and substance misuse pose serious risks to the fetus. Women suffering from either problem need help with their unresolved conflicting emotions of satisfying their own need with that of protecting their unborn child. Frequently they suffer from social isolation and a lack of understanding by their families and friends (Ho 1985; Merlin 1992; Laryea 1991). Women who are HIV positive tend to have multiple social problems and a high incidence of intravenous drug abuse (Johnstone et al 1992). They may worry about infecting their baby and be apprehensive about both future care of their baby and the possibility of their own untimely death.

The rate of risky health behaviour appears to be significantly related to a woman's internal belief concerning her control over pregnancy outcome. Work by Tinsley and colleagues indicates that this belief affects a woman's adherence to prenatal health care and in turn the number of periods of hospitalisation during pregnancy, as well as outcome of pregnancy. Drugs, smoking, and poor diet are related to low birth weight, premature labour and other negative fetal outcomes such as sluggish respiration at delivery. Social class is a factor, with those from the lower social classes more likely to attribute health and illness to chance than middle class women (Tinsley et al 1993).

Social class also affects both social support and information which, in turn, affects women's satisfaction with the quality of the birth experience. According to one study, women from lower social classes feel less well supported and less well informed than those of the middle classes (Quine et al 1993). Ley's work on communicating with patients shows that understanding and memory determine patient satisfaction with information. Failure to understand is, according to Ley, the result of three interrelated problems: the material is often too difficult for the individual to understand; the individual lacks elementary medical knowledge about their body;

the individual has active misconceptions that are so incorrect as to interfere with proper comprehension (Ley 1988). Ways of improving communication, content and style of information in relation to prenatal care, with particular attention as to how best to communicate with lower social class women, is felt to require further research (Quine et al 1993).

Pregnant disabled women

Disabled or chronically ill women can face, or perceive that they face, disapproval for having become pregnant (Rotheram 1989). Moreover, pregnancy may place an added strain on their physical condition as in maternal cystic fibrosis (Cohen et al 1980). Pregnancy can often restore confidence to a disabled or chronically ill woman as proof that she is feminine, has a body that can carry a pregnancy and produce a healthy child (Raphael-Leff 1991). Failure on the part of professionals to accommodate the handicapped in the delivery of antenatal care is a sobering criticism (Kelsall 1992).

Carty and colleagues describe the pregnant woman who is disabled as a "physically or sensory challenged individual," and the care provider's role as "challenging" (Carty et al 1990, p 133). They provide a constructive set of guidelines to assist the midwife who can then adopt the same positive approach to pregnancy as she would among able bodied women. For example, women with hearing impairment require an interpreter and the woman herself will need to decide if she is more comfortable with a woman, a friend, family member or professional interpreter (Carty 1990; Kelsall 1992). In relation to explaining prenatal tests it is important to speak directly to the woman herself and not to the interpreter. Women suffering from visual impairment do not gain from the printed materials provided either pre-pregnancy or during pregnancy. As a result they may have limited background knowledge about the anatomy and physiology of

their bodies, the changes to expect in pregnancy and the examinations and tests offered. The use of tactile models to aid explanation wherever possible is helpful (Carty 1990)

A guide to pregnancy and birth for women with disabilities emphasises that disabled pregnant women have the same concerns as able-bodied women and devote attention to how women with a disability feel about the idea of having a disabled child (Rogers and Matsumura 1991). The authors suggest that disabled women just as able bodied women need to ask themselves the questions: "How would having a disabled child change my life? How is this different from having an able-bodied child? Would it be possible to provide the extra care a disabled child might need?" (page 67).

Mental Health and Pregnancy

Studies have found psychological disturbance among pregnant women to be widespread. Anxiety, depression, worry and impaired concentration have been observed (Kumar and Robson 1984; Tunis and Golbus 1991). Between 10 per cent and 15 per cent of women have been shown to be depressed postnatally (Cox et al, 1982; Watson et al 1984) and a substantial number of these become depressed during pregnancy (Watson et al 1984). Mental illness associated with pregnancy and childbirth is reviewed comprehensively. (Kumar and Robson 1984; Brockington et al 1990).

Sharp (1988) in a longitudinal study of childbirth-related affective disorders used the General Health Questionnaire (GHQ) to screen a cohort of predominantly white British, working class women attending, for the first time, a South London antenatal clinic. Thirty five per cent of women scored positive on the GHQ at their first visit among whom 29 per cent were diagnosed as psychiatric 'cases', mainly neurotic depression. Both this study and others found factors such as unemployment, financial difficulties, housing, poor social support and poor marital relationship to be

associated with symptoms of stress (Paykel et al 1980). Although support of a woman's partner is recognised as significantly influencing the quality of both physical and emotional well-being of a woman during pregnancy (Brown et al 1986), the needs and vulnerability of men during pregnancy are being recognised and acknowledged (Raphael-Leff 1991). This comment was considered highly relevant to this study in view of the necessary participation of at least some male partners in the screening procedure.

4. 6 Men and pregnancy

Unlike other prenatal screening tests, the CF carrier test requires that the male partner be available for testing. If a woman is identified as a CF carrier it is of no consequence unless her partner is also a carrier. The information leaflet outlining the test and inviting women to be screened was sent with a woman's booking clinic appointment so that her partner was given the opportunity to read the leaflet and become involved from the outset. Prenatal CF carrier testing coincides quite incidentally with an increase in the participation of the father in all aspects of birth and childbearing. The researcher considered the degree to which fathers became involved in the decision to accept or decline CF carrier screening was an important aspect of the study and, therefore, experiences of men during pregnancy merited review.

Couvade syndrome

In traditional societies, men undergo rituals signifying their transition to fatherhood. Anthropologists have documented the ritual known as Couvade during which special observances and restrictions are performed (Bothamley 1990). Couvade is derived from the French word 'couver' meaning to breed or hatch. In some cultures the ritual entailed simulated childbirth by the father-to-be at the time of labour (Zalk 1980). Frequently there was abstinence from certain activities such as hunting, or dietary restrictions.

Couvade provided public recognition, social concern and most importantly an emotional outlet for the father-to-be (Raphael-Leff 1991). Psycho-somatic symptoms among expectant fathers are attributed to the underlying need to experience the pregnancy physically. Often the symptoms involve the alimentary tract; swellings, dental problems, backache, vomiting and weight gain are also documented (Trethowan 1968). The symptoms usually appear around the third month of pregnancy, are less severe in the middle, become more intense in the third trimester and disappear after delivery. The incidence is reported to range from around 10 per cent to 1 in 3 expectant fathers (Trethowan and Conlon 1965).

One study revealed up to 65 percent of prospective fathers developed symptoms resembling pregnancy (Shershevsky and Yarrow 1973). Other studies have looked at expectant fathers and a non-expectant control group and found no significant difference during pregnancy; however, in the immediate post-natal period expectant fathers manifested a significant difference in psychological symptoms (Clinton 1987; Gerzi and Berman 1981). In our modern society the past cultural and religious ceremonies which both marked and assisted coping with life transitions have all but disappeared. Nothing has replaced them and so individuals have to cope with transitional crisis alone (Blum 1980). It has been suggested that midwives and other health care professionals should offer anticipatory health counselling for expectant fathers (Clinton 1987).

Men, stress and pregnancy

The opportunity for men to voice difficulties in adjusting to pregnancy is often overlooked. "Society sanctions the symptoms experienced by women," but, " the male partner is expected to be the healthy provider and caretaker for his family " (Drake et al. 1988, p 438). Indeed, in one study men reported working longer hours

during pregnancy in response to anticipated financial expenditure or as a result of anxiety (Bothamley 1990) and in another, concerns about the risk of pregnancy and birth associated with a feeling of helplessness about, and responsibility for, the mother and fetus was voiced by men (Lewis 1986).

Nowadays the ultrasound scan allows the father early visual confirmation of the pregnancy. One study reported up to 84 per cent of partners attended the first ultrasound scan and found it a worthwhile experience. Most partners (68%) felt adequately prepared for the birth and ready to cope, but 22 per cent felt inadequate and unsure about what to expect (Pratt 1990). The conclusion drawn from this study was that a booklet should be designed which was devoted to fathers-to- be.

The pregnant woman's role is considered critical in involving the male partner "bringing her mate into the spotlight or keeping him in the wings" (Jordan 1990, p 14). Fathers in Jordan's study felt that health care providers viewed the mother and baby as their client and perceived his presence at prenatal visits as "cute or novel". They felt they were regarded only as in a supportive role to their partner and not treated as central to the proceedings.

A study which compared male and female psychological and emotional symptoms experienced during pregnancy found fewer than 15 per cent of women and only 10 per cent of men reported increased psychological well-being during pregnancy (Condon 1987). Marital insecurity was a significant fear among men who were concerned that "closeness" with their partner would suffer, or that their partner would become closer to the child. Men in this study also reported concern regarding future financial security and many were found to "work harder" (36%) and some "very much

harder". A significant finding was that multiparous couples reported a higher rate of psychological symptoms leading to the conclusion that the tendency to focus antenatal support on primiparous couples was a mistaken one.

It has been suggested that the sequence from pregnancy stress to parenting stress among pregnant women is important, more so than the stress of labour or delivery (Younger 1991). The identification of factors causing pregnancy related stress among men may well have similar implications. With regard to this study the researcher felt that any assessment of the impact of prenatal CF carrier screening should not focus entirely on the pregnant woman, but should include the male partner.

4.7 Questions emanating from the literature review

A lack of critical analysis of the literature reflected the potential difficulty of undertaking a study which was at the cutting edge of research. The researcher was unable to present a critical state-of-the-art summary of knowledge on a topic which had not previously been studied. There were no studies which directly addressed the impact of the offer of CF carrier screening on pregnant women. Despite this, the literature survey enabled the researcher to elicit a number of broad questions pertaining to cystic fibrosis screening from studies of previous genetic screening programmes among other ethnic groups; the psychological impact of being at risk of passing on a genetic disease to a child; prenatal screening and diagnosis; fetal or infant loss; pregnancy; and lastly, concurrent stressors.

Genetic screening

Screening should be voluntary, but will women who are offered carrier screening perceive that they have freedom of choice to accept or decline testing? Informed choice means being given clear accurate information tailored to suit the needs of the individuals

concerned, and thus enabling them to make a fitting decision. Pre-screening information and counselling is pivotal to informed decision making. Can the content and delivery of pre-screening information be tailored to suit the needs of the target population?

Pre-screening information and counselling is also crucial to ensure that the target population and those identified as carriers understand the harmlessness of being a single gene carrier. Can this be achieved?

Other genetic screening programmes have shown the benefits of community education in the uptake of screening and in avoiding public misconceptions about gene carriers. Where do women glean most of their information? Are they even familiar with the name "cystic fibrosis"?

The stage of life and the setting in which a test is offered also influences acceptability. Will women and their partners feel that an antenatal clinic is an acceptable environment to offer genetic screening, and pregnancy an acceptable time? From previous screening programmes it seems that pregnancy may be perceived both by the public and health care professionals as the optimum time to offer genetic screening. Will those who are identified as carriers agree?

Genetic Disease

Couples at risk of having a child with a genetic disorder have to come to terms with a series of losses. This loss is frequently combined with the need to assimilate information to enable them to make a decision regarding the continuation or termination of their pregnancy. How much information do these couples retain? Do they have any misconceptions ? Do they feel the information they received prior to screening and on

receiving their carrier test result met their particular requirements? On reflection do they regret having been screened?

Prenatal Screening and Diagnosis

Do women perceive the screening experience as stressful? The literature does not give an answer to this question. Will pregnant women's primary appraisal of CF carrier screening be that it is anxiety provoking or reassuring, or do they feel indifferent?

There is evidence that women who are identified as being at an increased risk of fetal abnormality through a screening programme find the experience particularly stressful. How will women who receive a positive CF carrier test react? Most studies have concentrated on measuring levels of anxiety but it is recognised that situations an individual deems as threatening generate feelings of anxiety and those experienced as loss generate depression (Brown et al 1988). What form intensity and duration of psychological reaction will emanate from being identified as a CF carrier during pregnancy? Can this be satisfactorily identified and measured? Do women adjust to their carrier state and can this be satisfactorily determined?

Which tests do women choose for their unborn baby? Are they selective about which screening tests they choose? What factors influence a woman's decision to decline or accept a voluntary screening test? Does comprehension of risk affect this decision? Are there socio-demographic factors which influence this decision?

Pregnancy

Pregnancy itself can be a time of conflict and stress. The CF carrier test is offered at the woman's first antenatal visit at a time when the majority of women are making adjustments to being pregnant and are experiencing the early emotional and physical

symptoms of pregnancy. Will the pregnant population of women find pregnancy an acceptable time to be screened? (This question is distinct from that of identified carriers who should be given the opportunity to reflect on whether pregnancy was the optimum time to be screened).

The support of the male partner is known to be an important factor in contributing to a woman's well-being during pregnancy. The CF carrier test differs from most other prenatal screening tests in its potential to involve the male partner. To what degree will male partners enter into the decision to accept or decline screening?

In attempting to assess stress in relation to a specific event it is necessary to take account of concurrent stressors. What percentage of the screened population will manifest signs of psychological disturbance at the time of screening? What are the major causes of concurrent stress among the pregnant women screened? Does concurrent stress influence a woman's reaction to being told she is a CF carrier?

The following chapter refines the questions and presents the research methods used to answer the questions.

CHAPTER 5

QUESTIONS AND SELECTION OF RESEARCH METHODOLOGY

Formulating the research questions

The opportunity to carry out this study arose from the researcher's involvement in a trial of prenatal carrier screening for cystic fibrosis presented by midwives in an antenatal booking clinic. One of the major questions which it was anticipated the trial would answer was: 'is this form of prenatal screening acceptable to women and their partners?' (page 32, table 2.4, question 4). It was this broad question that initiated a preliminary review of the literature and an overview of the screening trial. From this arose a number of key questions concerning the presentation of the screening test by midwives to the pregnant population and the care of those identified as carriers (page 48). A pivotal question revolved around the emotional impact of offering genetic carrier testing during pregnancy. This led the researcher to review the subject of stress and coping. A model of Stress, Coping and Outcome (Cochrane 1983) was used as a conceptual model for the study. The model helped delineate a number of areas which could affect the emotional response of women and their partners to prenatal CF carrier screening (table 3.1 page 64). A literature search of these areas clarified those questions which needed to be answered and these were refined in the context of Cochrane's model of stress and coping (Figure 5.1). The questions and areas of concern polarised around three discrete stages of the screening process: the pre-screening or threshold stage; the CF test result stage; and the post-screening result stage.

The Pre-screening stage

The literature search showed that previous knowledge; pre-screening information; perception of risk; reason for being screened; concurrent stress; demographic factors and current emotional and physical well-being could contribute to the uptake and impact of screening.

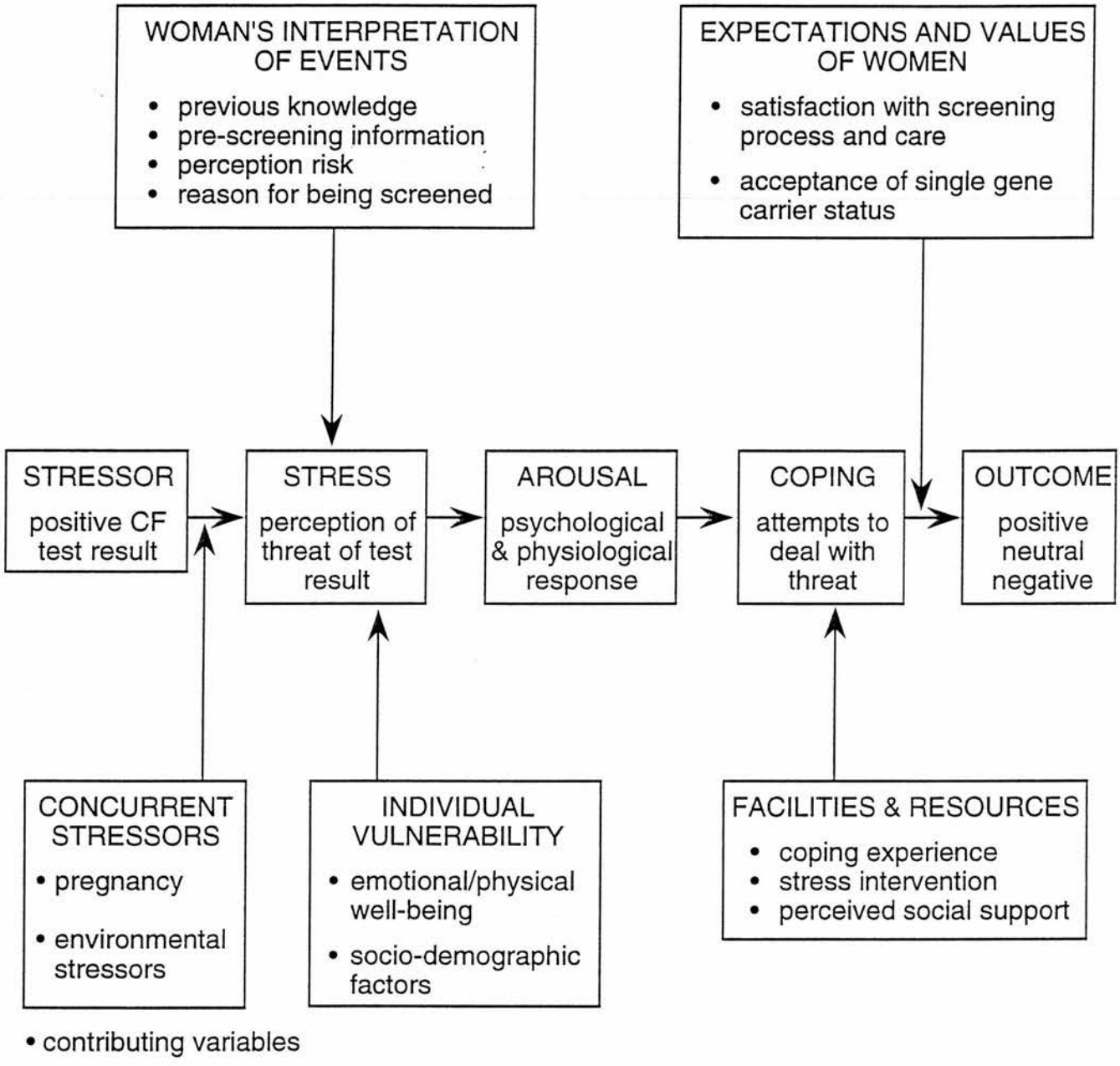


Figure 5.1. The application of Cochrane's model of stress and coping in formulating the research questions (adapted from Cochrane 1983).

The test result stage

The nature and level of a stressful response to receiving a positive test result would require to be assessed at the test result stage.

The post-screening result stage

The duration of a stressful response, acceptance of the single gene state and satisfaction with the screening process would require to be assessed after the partner's negative test result was known. Within the context of these three screening stages the questions were refined.

5.2 Research questions

1. **Are there pre-screening variables which influence a woman's response to prenatal CF screening?**

2. **What factors influence a woman to accept or decline prenatal CF carrier screening?**

3. **Will identifying a woman as a CF carrier during pregnancy provoke a stressful response both in her and her partner?**

4. **Do carriers and their partners understand the essential facts concerning CF carrier screening and what is their attitude toward having been screened?**

The overall aim was to assess the implications of prenatal genetic screening for midwifery practice.

5.3 Research methodology

A cohort of women who were offered CF carrier screening during the trial were studied. The study employed a descriptive, exploratory approach and was conducted prospectively using structured and semi-structured self-report methods.

5.4 Setting

Women were recruited from the antenatal clinic of the Simpson Memorial Maternity Pavilion in Edinburgh where a prenatal CF carrier screening pilot trial was in operation. During an 8 month period, from the beginning of May 1991 to the end of December 1991, women who received an invitation to enter the trial were recruited for this study.

5.5 Sample

Women were included in the sample if they met the following criteria:

1. They were English speaking
2. They were not more than 18 weeks gestation of pregnancy
3. They were in contact with their male partner
4. There was no abnormality of pregnancy detected at booking

Out of a total of 2,541 women 334 (13%) were not eligible to enter the study. Of 2207 eligible women 2058 (93%) participated.

5.6 Ethical Approval

Ethical approval was sought to carry out this study as an integral part of the screening trial from the Paediatric Reproductive Medicine Ethics of Medical Research Sub-Committee (Protocol 27/90). A standardised form was submitted on 1st June 1990 giving information on the significance of the study, confidentiality, informed consent

and the method to be used. Ethical committee approval was granted on 6th August 1990.

5.7 Access

The researcher met with the Director of Midwifery at the recruiting hospital and outlined the objectives, methodology, instruments and storage of data to be used during the study. Freedom of access to a woman's antenatal records was discussed. The researcher acquainted the antenatal clinic staff in the hospital with the nature of the study and comments and questions were invited.

5.8 Confidentiality and Informed Consent

Women were invited to participate in the study by means of an information leaflet outlining the trial. The leaflet (Appendix 1) was sent to them with their booking clinic appointment. The leaflet emphasised that one aim of the trial was to try to understand how women and their partners felt about the offer of cystic fibrosis carrier testing during pregnancy. A consent form was included in the leaflet which emphasised that a woman could withdraw from the study at any time. All data collected were stored on a dedicated computer and access was restricted to the researcher and the genetic nurse.

5.9 Constraints imposed by the study setting

The study setting imposed constraints on the choice of research methodology. Research methods which caused minimum disruption of patient care and intrusion on the midwife and pregnant woman were desirable. The researcher's schedule required to fit around that of the pregnant woman and the clinic staff caring for her. Research pertaining to carriers and their partners required to cause a minimum of intrusion on a couple who might be shocked or distressed. It was felt that lengthy interviewing would not be appropriate and could confuse a couple whose immediate need was that

of emotional support and counselling from the genetic nurse. Thus the researcher tried to balance the desire to obtain data for the study with the need to avoid disrupting midwifery care or intruding into the interaction of the genetic nurse with a couple. Continued follow-up of carriers and their partners needed to avoid carriers and their partners feeling over-researched or being continually reminded of a potentially traumatic episode in a pregnancy which was continuing normally.

5.10 Research instruments

The instruments chosen for answering the questions involved rating scales and specially designed questionnaires. These will be presented by addressing each research question in turn. All the questionnaires were submitted to a pre-test, to evaluate the adequacy of the tools in measuring the research variables, isolating bias, vagueness or inadequate questions as suggested by Barker (1991).

5.10.1 Question One:

Are there pre-screening variables which influence a woman's response to prenatal CF carrier screening?

The literature review of previous genetic and prenatal screening programmes indicated a number of variables which appeared to influence individual response in relation to both uptake of screening and psychological response to receiving a positive test result.

5.10.1.1 Variables measured:

1. Respondents' prior familiarity with the name cystic fibrosis:

Pre-screening information is known to be one important factor. Although an information leaflet will provide an introduction to screening, prior knowledge may be gleaned from the media or other sources which may influence understanding.

2. Women's opinion regarding the content of information in the pre-screening leaflet.

Women require information to help them to decide whether to accept or decline the invitation to be screened for CF during pregnancy. Criticism has been made of patient information leaflets for their tendency to be written in a format that health care professionals see as being generally applicable, rather than attempting to meet individual needs (Wilson-Barnett 1988). When the CF carrier screening leaflet was designed, it was deemed important to regularly monitor women's satisfaction with the content of information (Mennie et al 1992a) as this too could influence their whole perception of the screening test.

3. Involvement of the male partner in the decision to undergo screening.

Distinct from some other prenatal screening tests, the CF test has the potential to involve the male partner. Indeed availability of the male partner was a criterion for entry to the screening trial. It has been suggested that a majority of men regard the decision to accept or decline prenatal screening tests to be solely that of the woman (Sjogren 1992), therefore, the extent to which male partners became involved at the pre-screening stage was considered highly relevant to this study.

4. Respondents' perception of their risk of being a carrier:

Awareness of risk appears to determine, to some extent, women's emotional reaction to a positive screening test result. Women who enter screening programmes aware of their risk seem to find it less stressful than those who were unaware of their risk (Farrant 1985). Apart from pre-test education a decision to be screened may be influenced by a person's perception of the risk of being a carrier rather than the actual risk (Marteau et al 1991a; Lippman-Hand and Fraser 1978). Moreover, when an individual is confronted with the risk of genetic disease in their offspring they tend to be influenced more by their perceived ability to cope with an affected child than

by numerical risk. Thus they simplify the focus of concern which shifts from the probability of being at risk to that of being at risk and the potential to bear an affected child (Lippman- Hand and Fraser 1978). In turn a woman's knowledge and perception of the effects of the disorder may also influence her decision to be screened.

5. Respondents' emotional response to the offer of prenatal CF carrier screening:

A woman's awareness of a particular fetal abnormality is likely to increase with the advent of a screening test. Some suggest that the very existence of a screening test creates anxiety in a woman by alerting her to a condition about which she was previously unaware (Rothman 1988). Yet, the lack of research means reserving conclusions about whether screening is inherently stress provoking (Marteau and Slack 1992a). A woman's initial response to the offer of screening might influence her decision to accept or decline the test and was thought a likely indicator of how she would feel until her test result was known.

5.10.1.2 Research Instrument

A self - administered questionnaire called the pre-screening questionnaire was designed to measure these five variables. The front cover of the questionnaire repeated the aims of the study as outlined in the screening information leaflet. Care was taken to comply with guidelines outlined in the literature which would assist in a high return (Crossfield 1988; Rees 1990; Basford 1990). These were that the questionnaire should aim for simplicity and clarity both in design and content and be kept as brief as possible. That questions should be arranged in a logical sequence to allow the respondent to develop thought processes and that they should not be loaded or biased so as to avoid achieving a preconceived result. Finally a pilot trial should be carried out on the target population to assess the usefulness of the questionnaire and identify any problems. A pilot trial of 167 patients revealed that 25 (15%) patients did not know

their risk of being a CF carrier and because this option had not been made available to them they had written "don't know" on the questionnaire. Question six was subsequently amended to include this option. The questionnaire was finalised and functional at the beginning of May 1991 (see appendix).

Demographic data

Age, socio-economic background, marital status and parity may influence a respondents' attitude to screening, comprehension of information, and in turn influence the decision to accept or decline a test. Marital status was considered an important variable because CF carrier screening required that the male partner be both available and willing to be screened if the woman was identified as a CF carrier. However, it was acknowledged that an available and supportive male partner was not necessarily associated with marital status.

Younger women and those with a higher level of education showed less inclination to make use of prenatal screening (Tymstra et al 1991). In addition, educational background of the male partner has been shown to influence a couples' comprehension of genetic information (Emery et al 1979). During the booking procedure the midwife collected an amount of demographic information and this was recorded on a woman's antenatal liaison card. A demographic information form was designed and completed for each respondent from data recorded on her antenatal liaison card. Area of residence was recorded by a numeric code; social class was assessed from the occupation of the head of the household (Office of Population Censuses and Surveys 1980); and a woman's decision to accept or decline MSAFP screening was also noted. Educational background data was forfeited because of appearing intrusive. Moreover, if screening were to continue after the trial then patient assessment by the midwife would be limited to information already recorded during the booking-in procedure.

5.10.2 Question Two:

What factors influence a woman to accept or decline prenatal CF carrier screening?

Why women choose a specific prenatal test is not entirely clear. There are no large studies which have addressed this issue. Are women selective about the tests they choose for their unborn baby or do they simply accept what is offered? A closed question inviting respondents to select from pre-assigned categories of response was considered. Such a method would have relied upon the researcher's interpretation of the phenomenon rather than the subjects' interpretation. There were no grounds to believe that a list of reasons why women might wish CF carrier screening in pregnancy as perceived by a health care professional would truly reflect those of pregnant women.

5.10.2.1 Qualitative Research Methodology

The fundamental characteristic of qualitative research is that it aims to take the subject's perspective on a subject and has been described as 'seeing through the eyes of the people being studied' (Bryman 1990). Qualitative research requires a minimum of structure and research involvement as it aims to emphasise the perceptions and subjectivity of the individual. The researcher tries to learn the experience of study participants and to understand their perspective of a situation (Polit and Hungler 1989).

If a research topic lacks previous information, qualitative research methodology has particular strengths which are lacking in quantitative methods of research (Field and Morse 1990). In particular, quantitative research requires the researcher to make theoretical assumptions whereas qualitative research does not. Thus, if an extensive library search reveals very little information about a topic, Field suggests it is probably not sufficiently developed to use quantitative research methods. Moreover, some assert that there is a need for nurse researchers to generate nursing theory within nursing

contexts rather than verifying theories developed outwith their own domain ((Duffy 1985).

Since the 1960's there has been a debate about these two major approaches to research (Bryman 1990). Frequently researchers restrict their choice to one or other (Field and Morse 1990), however, some suggest that when quantitative and qualitative research are jointly pursued a more comprehensive account of social reality emerges (Bryman 1990). Combining both research methods has been used to further nursing knowledge. Hockey (1976) used structured interviews and incorporated open-ended questions to collect data for a study entitled 'Women in Nursing.' Here, use was made of a quantitative research tool to measure nurses' job satisfaction. By linking interview comments to the respective scores, Hockey was able to build a profile of nurses in relation to job satisfaction. More recently, a major National Health Service (NHS) Survey, involving nearly thirty eight thousand NHS staff, combined both methods most effectively. The study was designed to glean insight into both knowledge and impact on staff of NHS reforms. The qualitative research study, based upon subjects comments, revealed a rich source of information about individual perceptions and anxieties regarding NHS reforms. Quantitative research methodology would not have demonstrated the emotional impact inflicted upon NHS staff by a major shift in organisation (The National Health Service Scotland 1992).

Women telling of their experiences of prenatal diagnosis has given insight into how one simple procedure can completely change a woman's experience of pregnancy (Rothman 1988). Women report being overjoyed by being pregnant but within a short time were faced with a test which led them into an ethical conundrum of astounding proportions. Rothman interviewed more than 120 women who were at risk of having a child with a birth defect. Around half the women had accepted the offer of an amniocentesis and the

remainder had declined intervention. Much of the interview data were collected by postal questionnaire but women wrote vividly of their experience. Notably this study revealed the variety of ways that women perceive this one procedure. It is the differing perception of the meaning of a prenatal test which requires to be taken into account when counselling women for prenatal screening and diagnosis. It was this diversity of perception among women which the researcher wished to expose and examine.

5.10.2.2 Method of collecting data

Question 9 of the pre-screening questionnaire asked women 'to say in a few of their own words' why they came to their particular screening decision. Although a qualitative approach was used to obtain the data it was then analysed quantitatively.

Analysis of speech or written text demonstrates that an idea can be expressed in many ways. The key word that actually defines it may not actually be used, thus an individual may state they wish 'more information' yet never use the term 'information' choosing instead: 'know more' or 'more facts'. This means the text must be examined for topics rather than linguistic content and coded accordingly (Tesch 1991). A computer database manager can efficiently store and retrieve information by searching across stored records (Fielding and Lee 1991). For the purposes of this study the 'dBase III plus' database management programme was chosen to store and analyse subjects' quotations (Tsu-der Chou 1986). The programme which was in general use within the researcher's department permitted two research colleagues familiar with the programme access to the data for the purpose of 'peer examination' (Field and Morse 1990). This involved soliciting help from colleagues in examining data to see if they can recognise the same topics and categories as those identified by the researcher and is a means of ensuring the credibility of the data and avoiding potential bias.

5.10.3 Question Three:

Will identifying a woman as a CF carrier during pregnancy provoke a stressful response both in her and in her partner?

5.10.3.1 Variables measured

Cochrane's model of stress, coping and outcome recognises the importance of individual perception of an event . The way in which an individual interprets a situation is the key to determining whether or not they regard a situation as stressful. Clearly an individual's own frame of reference is crucial to his or her initial attitude and is likely to influence their response if the test result is positive. Thus different individuals may react uniquely to a similar situation which is dependent upon the degree of perceived threat (Cochrane 1983). Pregnancy itself is viewed as a psycho-biological crisis (Blumberg 1984) and therefore psychological response to pregnancy needs to be taken into account. A further consideration is introduced in Cochrane's model of stress and coping which emphasises the importance of recognising other provoking agents in an individual's life. Cochrane states that these too must be considered when assessing an individual's response to a stressful situation (Cochrane 1983). The criteria for selecting an instrument was that it could detect a stressful response in an individual, identify the nature of their response and measure the degree and duration of their response.

5.10.3.2 Research Instruments

The General Health Questionnaire

The General Health Questionnaire (GHQ) is a self-report instrument designed to detect current short-term psychological disturbance of a non-psychotic nature (Goldberg and Williams 1988). It identifies two main classes of problem: inability to carry out one's normal 'healthy' functions and the appearance of new phenomena of a distressing nature. Although originally developed for use in general practice settings, the GHQ has

been used in both hospital and community settings in numerous surveys which indicate it is suitable for use with men and women of all age groups in detecting psychological distress (Thornley et al 1991; Maskey 1991; Gage and Leidy 1991; Stoate 1989; Goldberg and Williams 1988). It has also been used with a multi-cultural sample of mothers with young children (Watson and Evans 1986). The GHQ was designed to cover four identifiable elements of distress: anxiety, depression, social impairment and organic symptoms. An individual with a positive GHQ score might be said to be emotionally stirred up and altered in respect from his or her normal self (McDowell and Newell 1987). The GHQ screens for acute rather than chronic distress, and was meant to be a first-stage screening instrument, with emphasis being on changes in condition not on the absolute level of the problem. Items compare the person's present state to their normal situation with responses ranging from 'less than usual' to 'much more than usual' (see appendix). The GHQ is a screening tool which is used to identify individuals who then require more extensive examination in order to ascertain the cause and nature of their psychological disturbance (Goldberg and Williams 1988). Items on the GHQ can be scored using conventional 0-1-2-3 Likert scores, but Goldberg recommends a simpler system of a two-point score, rating problems as present or absent and ignoring frequency (Goldberg and Hillier 1979). Thus in the original 60-item version of the questionnaire any 12 positive answers constituted a positive GHQ score. Such an individual would then be interviewed to determine the nature and cause of their response.

There are five versions of the GHQ ranging from the 60-item down to a 12-item questionnaire. Given that time is at a premium in an antenatal booking clinic the shortest version, the 12-item GHQ, which takes one and a half to two minutes to complete, was selected (Goldberg and Williams 1988). A cut off point of 3 (i.e. 3 or more is a case) has been tested for the GHQ-12 in males and females (Nott and Cutts

1982; Banks 1983; Mari and Williams 1985). On the basis of an extensive review Goldberg and Williams (1988) concluded that the validity of the GHQ-12 is comparable with that of the longer versions. The 12-item GHQ has been used in a number of studies (Banks 1983; Briscoe 1989; McGrath et al 1989; Stafford et al 1980) and found acceptable to both males and females in detecting stress. Critical to this study was its previous use among populations of women during pregnancy and in the immediate postnatal period (Sharp 1988; Briscoe 1989; Nott and Cutts 1982). The 30-item GHQ was validated in pregnancy by Sharp (1988). At their first antenatal visit Sharp asked 179 women to complete the measure and 35 per cent were found to be high scorers.

Briscoe found the instrument was acceptable to mothers and was particularly useful in helping them to express their feelings. "Behind a smiling facade" she found women who were severely depressed and guilt ridden. Such women seemed to find it easier to admit their feelings in a pencil and paper test, a conclusion found in other studies of pregnant women (Jones 1990). This reluctance to admit and seek help for emotional problems is also confirmed by Nott and Cutts (1982). The 28-item GHQ has recently been used to assess pregnant teenagers at 'booking' for psychological disturbance (Maskey 1991).

Goldberg has summarised data on the association between the GHQ and demographic variables. Females tend to show higher scores; there was no association between age and GHQ scores; there was a significant tendency for lower social class respondents to have higher scores. The Manual of the General Health Questionnaire gives the optimum cut-off point for the 12-item GHQ as 2 or 3 but emphasises the need to recalibrate the measure in a representative sample of the population for which it is intended. Therefore, a cohort of 167 antenatal patients and 77 male partners were asked to complete a 12-item GHQ at their first antenatal visit. Using a

cut off point of 2 items or more, resulted in 51 per cent of women scoring positive and 18 per cent of males. By raising the cut off point to 3 (3 items or more is a high score), lowered the prevalence of high scorers to 31 per cent in females a figure compatible with Sharp's study of antenatal patients, and 10 per cent in males (Sharp 1988). It was considered feasible to carry out more intensive investigations on around thirty percent of women at the antenatal clinic but considered impractical to do so on fifty per cent, therefore, a cut off point of 3 was chosen for this study.

The Symptom Rating Test (SRT)

The GHQ will identify cases of stress but it is not designed to measure symptoms and symptom severity although it does monitor change (Goldberg and Williams 1988). The Symptom Rating Test (SRT) (Kellner and Sheffield 1973) was developed to measure distress. A self-report questionnaire, the SRT is based on a checklist of 30 symptoms, 7 somatic and 23 psychological (see appendix). These symptoms are then self-rated for severity. Four subscales are included: anxiety, depression, inadequacy and somatic concern. It has been used in a number of drug trials because of its great sensitivity to change (Henderson 1982). It was considered unwise to ignore the potential existence of depressive symptoms in both the pre-screening and post-screening phases of the CF carrier testing procedure.

Studies have found psychological disturbance among pregnant women to be widespread. Anxiety, depression, worry and impaired concentration having been observed (Kumar and Robinson 1984; Tunis and Golbus 1991). Other studies report that the incidence of depression rises significantly in early pregnancy and in the first three months after delivery (10 per cent and 14 per cent respectively) (Cox et al 1982; Watson et al 1984). To identify subjects suffering from depressive symptoms was calculated to be of importance not only to the findings of the study, but of critical

importance to a woman's well-being during pregnancy and in the post-natal period. To focus only on symptoms of anxiety as an outcome of receiving a positive screening test result was considered unwise because depression was regarded as a possible manifestation of stress in relation to loss (Brown et al 1988). The SRT measured both psychological states and in addition could detect change in feelings of inadequacy and somatic symptoms. Unlike the GHQ the SRT was not designed as a case-finding instrument (Thomson 1989) but to measure distress in such a way as to be able to detect changes in the state of the individual (Kellner and Sheffield 1973). It has been used in a number of epidemiological surveys which include psychological disturbance as a variable. The questionnaire differentiates sensitively between levels of distress in different groups (Kellner et al 1984; Magni et al 1982). It has been used to measure psychological disturbance in women undergoing amniocentesis and was found to measure sensitively changes in distress (Fava et al 1983).

In the present study, the value of the SRT was its ability to detect symptoms of stress which could be identified and measured for both degree and duration. There are 4 possible answers to each SRT question: not at all (score =0), slightly (score=1), a great deal, quite a bit (score =2), extremely, could not have been worse (score=3). Studies using the SRT have shown average total distress scores of 8 to 11 for the general population, and 49 for those with symptoms of psychological disturbance. The SRT takes 2 minutes to complete and the 30 symptoms are defined in simple every day language so as to be easily understood and non-threatening to the respondent (Cochrane 1980). Incorporated in the pre-screening questionnaire was a 12-item GHQ designed to assess the threshold emotional status of a woman prior to being screened. The Symptom Rating Test (SRT) was used to identify the nature of distress among women who presented at the clinic with a positive GHQ score.

5.4.4 Question Four

How well do carriers and their partners understand the essential facts concerning CF carrier screening and what is their attitude toward having been screened?

Follow-up of individuals who have received genetic counselling has been recommended (WHO 1969) as a valuable complement to genetic counselling. Only by assessment of carrier's and partners knowledge and comprehension is it possible to learn how well they understood the counselling information given to them. Moreover, insights gleaned from recipients of screening may identify dissatisfactions leading to improvements in the delivery of screening (Reynolds et al 1974).

5.10.4.1 Research instrument

A self report questionnaire was designed consisting of closed questions called the 'Facts and Feelings Questionnaire'. The 'facts' section was composed of six statements derived entirely from the pre-screening information leaflet and corresponded in sequence to the information given in the leaflet. Respondents were asked to tick which statements they thought to be true. Participants' attitudes toward the prenatal screening trial specifically and towards CF carrier testing in general were assessed in a section of the questionnaire entitled the 'feelings' section. A pilot trial tested the questionnaire among 20 screened women and their partners. The initial lay-out of the questionnaire caused some difficulty. The lay-out of the questionnaire was altered but the questions themselves remained unchanged. The questionnaire was piloted for a second time and was found by respondents to be simple and quick to complete. A computer data base was designed for storage and analysis of the questionnaire data.

An outline of the study protocol is shown in figure 5.2

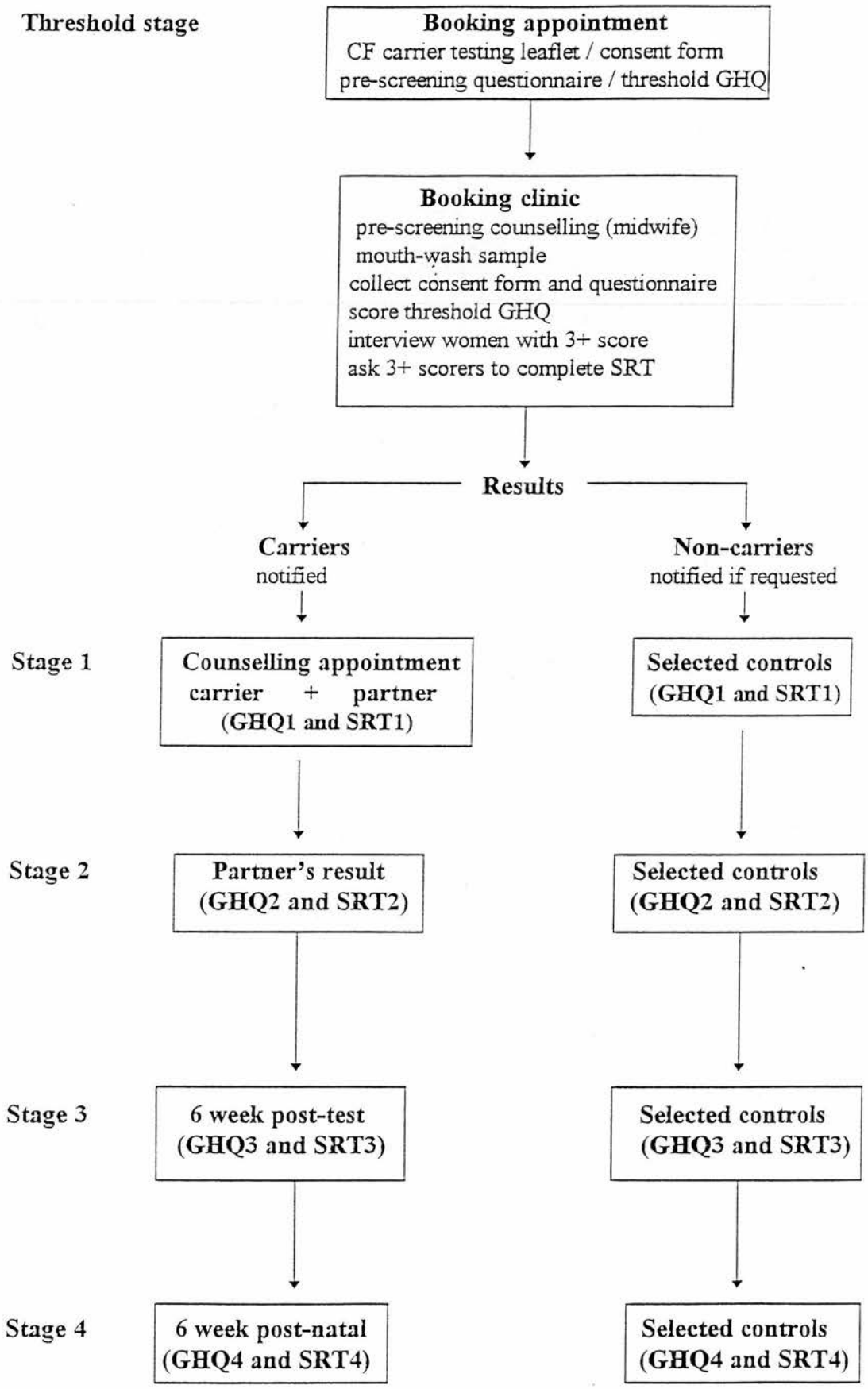


Figure 5.2 Study Protocol

5.11 Study protocol

Women were sent a pre-screening questionnaire incorporating a GHQ (termed threshold GHQ) with the leaflet inviting them to participate in the screening trial. They were asked to complete the questionnaire and bring it to the clinic. Women entering the trial who had not completed a questionnaire were asked to complete one at the clinic. Signed consent was obtained from women entering the study. The GHQ was scored by the researcher. Those with a positive score (3 items or more) were interviewed to establish the likely reason for their response and asked to complete a SRT (termed threshold SRT). GHQ and SRT scores along with interview data were recorded on a computer database for recall when a carrier was identified. Women identified as carriers were contacted a week later by telephone or, in a minority of cases, by letter and invited to attend the hospital, along with their partner, for genetic counselling. The couple were seen prior to genetic counselling by the researcher and the aims and sequence of the questionnaires explained. Male partners were asked to sign a consent form and both partners were asked to complete a GHQ and a SRT (termed GHQ1 and SRT1)

On receipt of the partner's negative test result the genetic nurse contacted the couple in all cases by telephone and informed them of the result. A letter was sent confirming the partner's negative result. Enclosed was a stamped addressed envelope and a GHQ and a SRT (termed GHQ2 and SRT2). Six weeks later the couple were sent a further postal GHQ and SRT (termed GHQ3 and SRT3) and a Facts and Feelings Questionnaire. Finally six weeks after the delivery of their baby the same two psychological measures were sent (termed GHQ4 and SRT4). Controls were contacted by telephone in all but 4 cases where contact was made by letter. Control couples received a postal GHQ and a SRT at comparable intervals to carriers and partners.

The results of the research are presented in the following chapter.

CHAPTER 6

PRESENTATION AND ANALYSIS OF DATA

The results of the study will be presented by addressing each research question in turn.

6.1 Question one: Are there pre-screening variables which influence a woman's response to prenatal CF carrier screening?

6.1.1 Sample and method

A pre-screening questionnaire was sent along with the CF carrier test information leaflet to 2,541 women who received an antenatal booking clinic appointment between May and the end of December 1991. A total of 334 (13%) women did not meet the criteria for entry to the study for reasons of late gestation of pregnancy, abnormality of pregnancy, unavailability of the male partner or poor command of English. Of 2207 eligible women 2058 (93%) returned a questionnaire. Questionnaires were completed either in the woman's home or in the antenatal booking clinic.

6.1.2 Presentation of data

The results for each variable measured will be presented in turn.

6.1.2.1 Characteristics of the study population

The characteristics of the study population are summarised in table 6.1. Significance of differences in characteristics was evaluated by the Chi squared test.

Analysis of subjects' age and social class revealed that there was a significantly higher proportion of young women (16 to 25 years) among socio-economic groups 4 and 5 and the unemployed (table 6.2)

Table 6.1 Characteristics of the study population n = 2058
(numbers and percentages)

Age (years)		
mean	28 ± 5·4	
range	16 - 44	
Parity		
0+0	804	39%
0+1 >	226	11%
1+0 >	1028	50%
Gestation		
mean	12·31 ± 2·35	
range	6 - 18	
Marital status		
married	1515	74%
single	458	22%
divorced	53	3%
separated	29	1%
widowed	3	0.1%
Social class		
1	262	12%
2	633	31%
3	674	33%
4	183	9%
5	105	5%
unemployed (ue)	163	8%
student (st)	38	2%
Religion		
Protestant	1128	55%
Roman Catholic	292	14%
Christian	60	3%
Other	57	3%
None	521	25%
MSAFP screening		
accepted	1918	93%
declined	140	7%

Table 6.2. Percentage of subjects in each socio-economic group; by age

	socio-economic group								
age	1 %	2 %	3 %	4 %	5 %	U/E %	stud %	n	p
16-20	1	5	30	18	11	32	3	146	<0.001
21-25	3	16	40	15	11	12	3	425	<0.001
26-30	13	34	35	7	4	5	2	846	NS
31-35	20	41	27	6	2	3	1	490	NS
36-44	27	40	24	3	1·5	3	1·5	151	NS

Analysis of subjects' age with marital status showed that younger women 16 to 25 years were significantly less likely to be married (table 6.3).

Table 6.3. Percentage of subjects in each marital group; by age

	marital group					
age	married %	single %	sep/div %	widow %	n 2058	p
16-20	11	88	0·5	0·5	146	<0.001
21-25	56	41	3	0	425	<0.001
26-30	84	12	4	0	846	NS
31-35	87	8	5	0	490	NS
36-44	85	11	3	1	151	NS

6.1.2.2. Prior information about cystic fibrosis

Eighty five per cent of subjects claimed to have heard of CF before reading the pre-screening leaflet. Television was the medium most frequently cited as their source of information about CF (Figure 6.1).

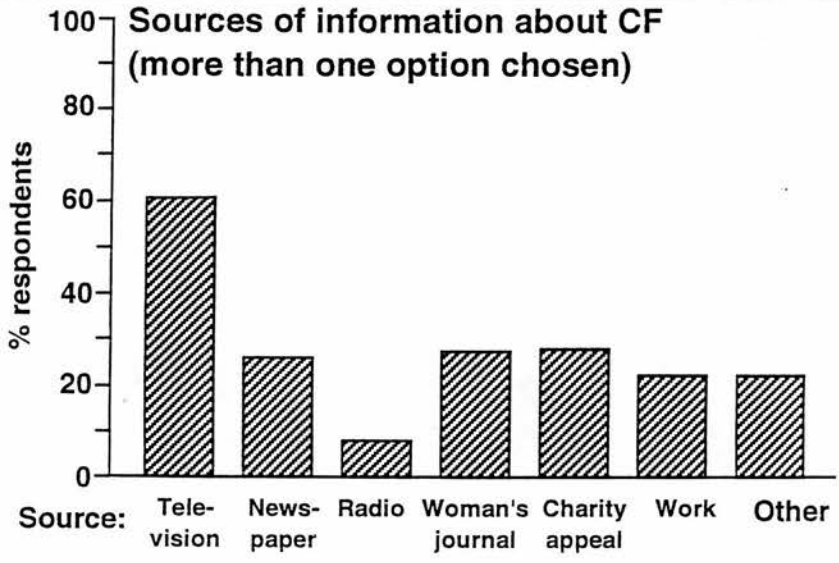


Figure 6.1 Sources of information about cystic fibrosis

Women belonging to socio-economic groups 1 and 2 were more likely to cite radio and newspapers as a source of information than women from other groups. However, almost equal numbers of women from all socio-economic groups had derived information from women's journals and charity appeals. Of those who indicated that they had heard of CF through their work, almost 70 per cent also belonged to socio-economic groups 1 and 2.

Young women (16-20 years) were significantly less likely ($p = <0{\cdot}001$) to have previously heard of CF than women who were older (Figure 6.2).

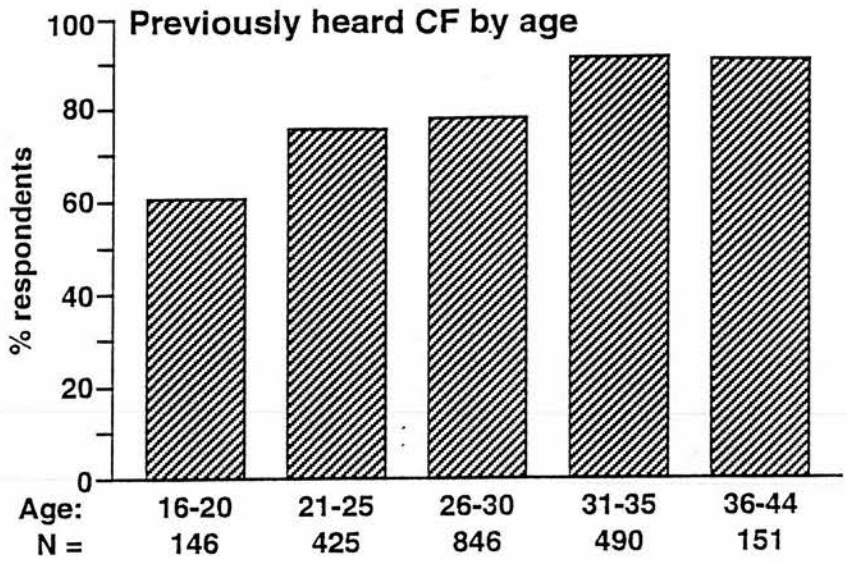


Figure 6.2. Women who had previously heard of CF; by age

6.1.2.3 Perceived understanding of the pre-screening leaflet

A majority of respondents (94%) stated they had found the leaflet easy to understand. Women belonging to the younger age groups (16-20 years and 21-25 years) were significantly more likely ($p = <0{\cdot}001$ and $p = < 0{\cdot}005$ respectively) to state that they had found the leaflet difficult to understand (Figure 6.3).

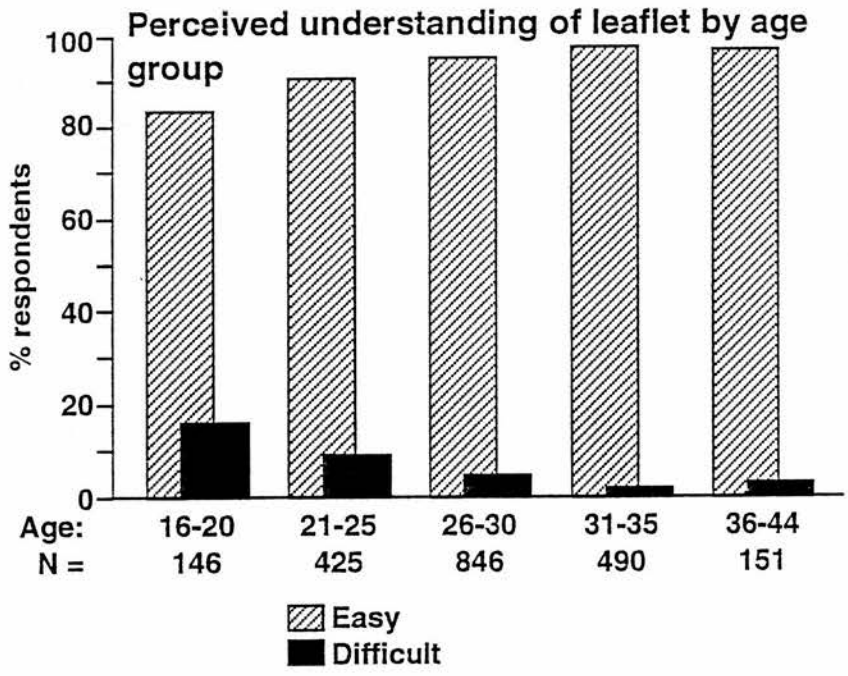


Figure 6.3. Women's perceived understanding of the leaflet; by age

6.1.2.4 Male partners who read the pre-screening leaflet

A majority of male partners (1438, 69%) read the leaflet. Of these 94 per cent were claimed to have found it easy to understand. Younger women (16-20 years) were significantly less likely (p = <0·001) to have a partner who had read the leaflet (Figure 6.4).

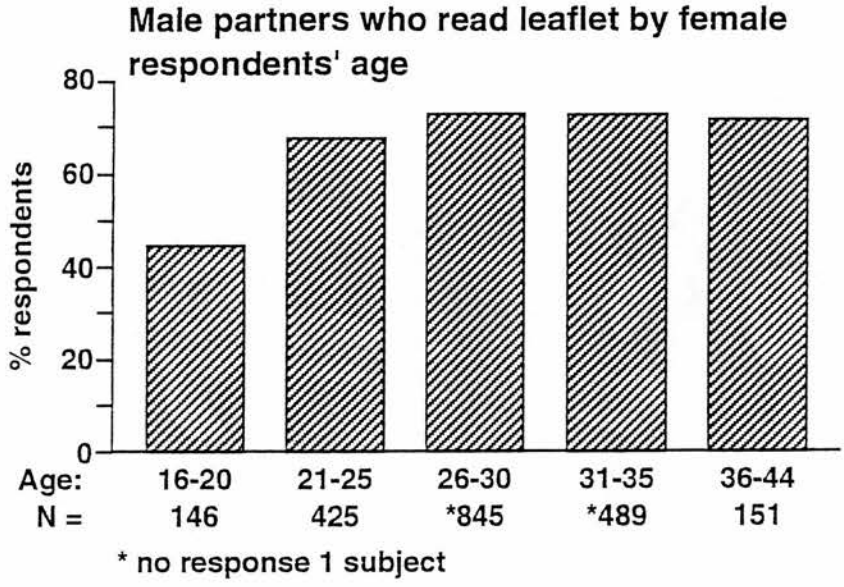


Figure 6.4 Male partners who read the leaflet; by women's age.

Younger women (16-20 years and 21-25 years) were significantly less likely o have discussed the test with their partner (p = <0·001 and p = <0·005 respectively). A significantly greater number of this age group were unmarried (Table 6.3) and this is reflected in the fact that significantly more married women (75%) than single women (51%) had discussed the test with their partner (p = <0·001) (Figure 6.5). However, single women were more likely, though not significantly so, to discuss the test with a significant other person, usually a relative (Figure 6.5).

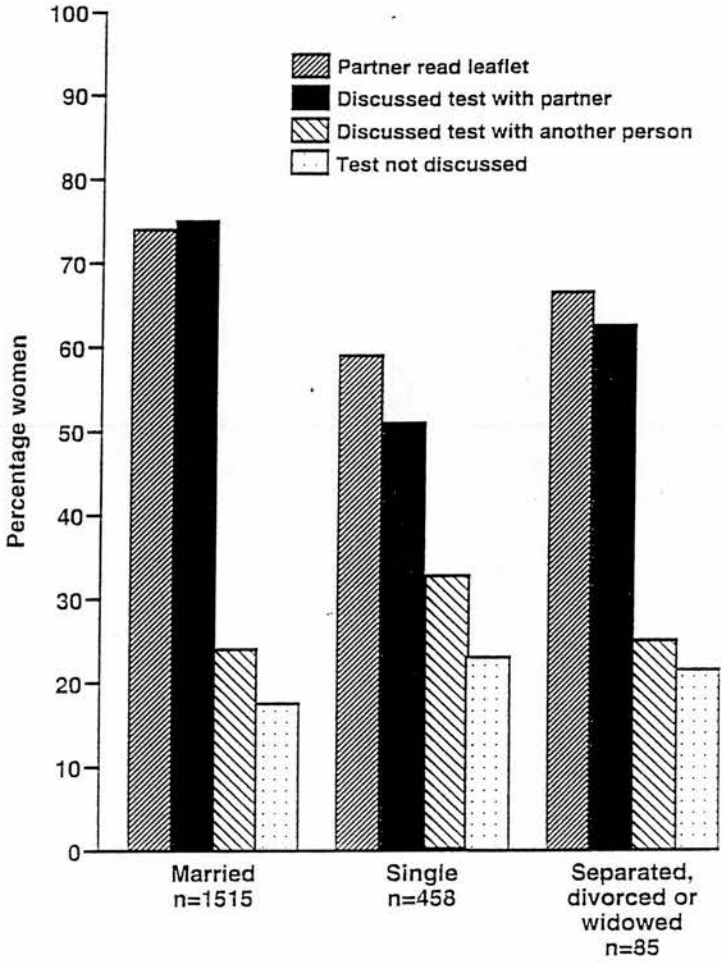


Figure 6.5. Involvement of male partner and significant others in screening decision; by marital group

Most women who were separated or divorced had become pregnant through a new relationship. A majority of their partners had read the leaflet and most had discussed the test with him (Figure 6.5). Three women who were widowed had new partners all of whom had read the leaflet and each of these women had discussed the test with their partner and a significant other person. Fifteen per cent of married women and 20 per cent of single, separated or divorced women claimed not to have discussed the test with anyone other than the midwife or the genetic nurse at the booking clinic (Figure 6.5). Among this group are an unestimated but probably small number of women who failed to receive an information leaflet prior to the booking clinic. A more substantial number chose not to read the leaflet prior to the clinic.

Only 50 women, out of the total population of 2,058, had discussed the test with their general practitioner and just 9 women discussed the test with their health visitor.

6.1.2.5 Women's perception of their carrier risk.

Most women (58%) knew that they had a 1 in 25 chance of being a carrier of a single CF gene (Table 6.4).

Table 6.4 Women's understanding of their carrier risk

		Risk options			
1 in 4 %	**1 in 25 %**	**1 in 100 %**	**1 in 200 %**	**don't know %**	**number respondents**
18	58	1	1	22	2058

Women in the younger age groups (16-21 years and 21-25 years) were significantly less likely ($p = <0{\cdot}001$) to accurately know their carrier risk (Figure 6.6).

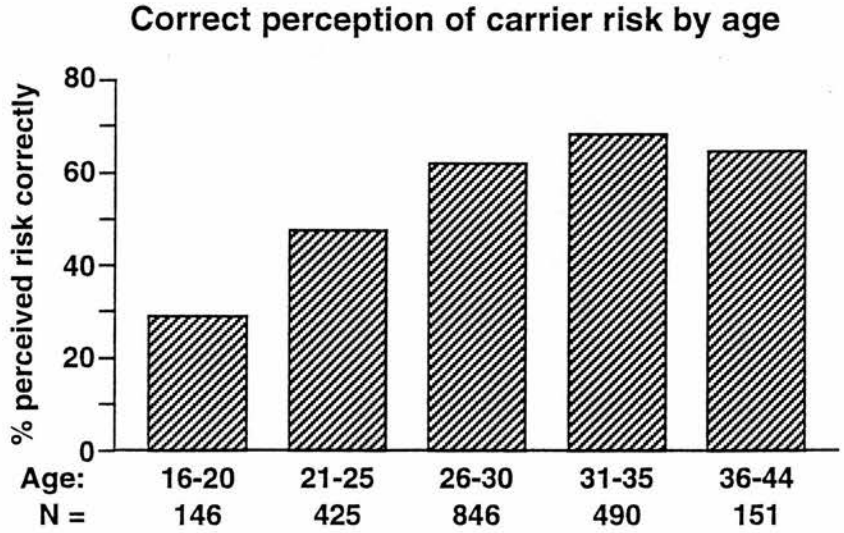


Figure 6.6 Women who understood their carrier risk correctly; by age

6.1.2.6 Emotional response to screening

Forty three per cent of women stated that they felt reassured by the invitation to be tested, while 25 per cent felt anxious. A total of 31 per cent indicated that they did not know how they felt emotionally toward being screened (Figure 6.7).

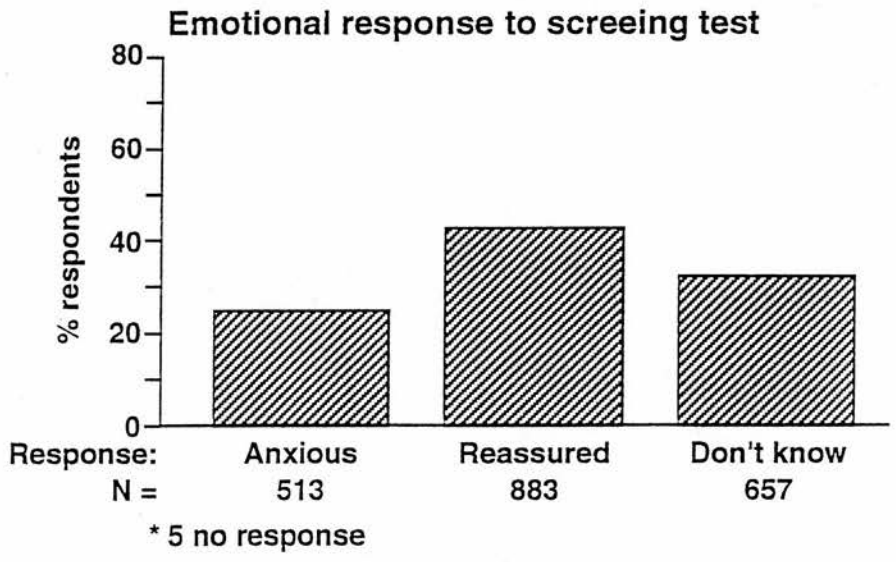


Figure 6.7 Women's emotional response to screening

There was no correlation between either a woman's age or knowledge of carrier risk and emotional response to screening.

6.1.2.7 Additional information requested in the leaflet

More information was requested by 644 (31%) of women: principally about diagnosing CF prenatally in the fetus; and the disease and treatment in a child born with the disorder (Figure 6.8).

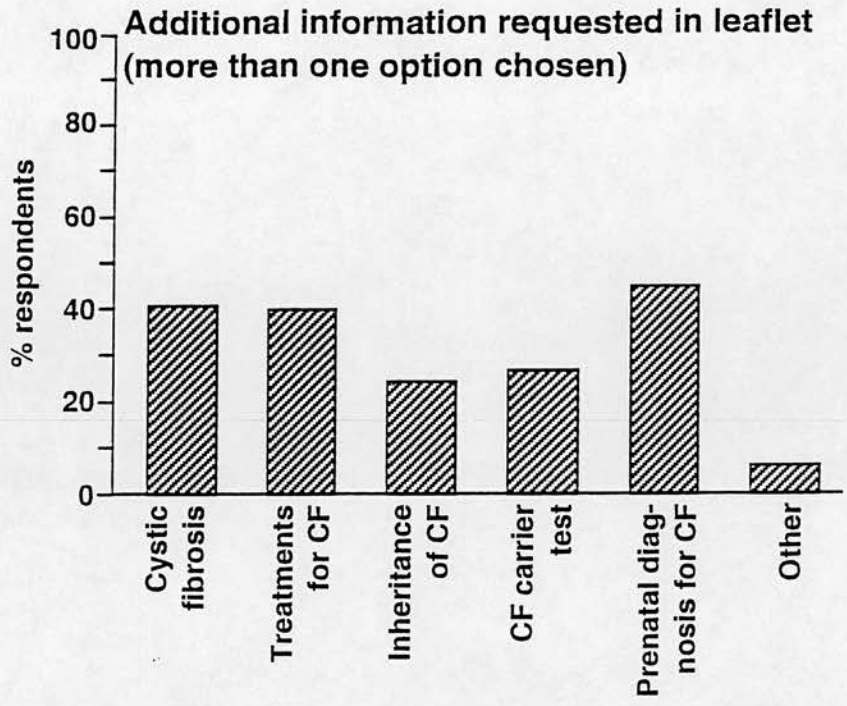


Figure 6.8. Topics of additional information requested in the leaflet.

Those who wished further information about prenatal diagnosis of CF were occasionally concerned about the procedure:

"I'd like to know how the unborn child is tested and the risk of possible miscarriage as a result of the test."

The majority of women who felt that the leaflet had provided them with sufficient information made comments such as: *"everything understandable"* or *"brief and clear"*

Others commented in a similar fashion to the following women:

"I'd only want to know more if both myself and my partner were identified as CF carriers."

Another concern voiced was:

"Would you be sure (100%) that an unborn child had CF or is there a chance a healthy child could be terminated."

Almost as many women (41%) wished to know more about the disease CF (Figure 6.8).

"I believe CF can vary in severity and would have liked to know more regarding this."

Forty per cent of those who requested further information in the leaflet requested more facts about treatments for the disease:

"More information on the amount of medical care needed by babies with CF."

The likelihood of curing the disease was also a concern:

"We would like to know about progress being made with research towards a cure."

Twenty three (4%) women requested 'other information' to be included in the leaflet. They often requested more detailed information.

"Statistics relating to % of babies born with CF. Further statistics on range of life expectancy."

Some offered constructive suggestions:

"The booklet mentions the rather alarming statistic of a 1 in 25 chance of being a carrier and a 1 in 4 chance of a baby having CF if the 2 recessive genes meet. I do not see a statistic to show how many babies (per 1000 for example) are born with CF. I feel this statistic might well reduce any alarm caused by seeing a 1 in 25 carrier risk."

Others had clearly misinterpreted the information outlined in the leaflet:

"If this is not a hereditary disease, then what causes some people to carry the disease? gene? - diet? - living standards? etc."

and one woman beseeched:

"What causes the disease? What hope of help if I had it? What hope of help if the child had it?"

6.1.3 Discussion of results

The response to the questionnaire was very high with 93 per cent of women completing a questionnaire. Women who failed to complete a questionnaire prior to the clinic were seen by the researcher who explained the aims of the study. Many were pleased to complete the questionnaire at the clinic: *"It will help pass the time"* was a frequent comment. The majority of women completed a questionnaire at home but an unestimated minority completed it at the booking clinic before discussing the test with the midwife. It could be argued that the environment in which the questionnaire was completed would reflect upon the results. It was felt that this was more likely to affect the outcome of psychological testing, the results of which will be discussed later in this chapter.

There was a social class bias toward the upper socio-economic groups (table 6.1) which may reflect both the fact that women from these groups tend to book earlier (before 18 weeks gestation) (Lester and Farrow 1988) and that a considerable group from the lower socio-economic groups booked at another clinic. In addition, it may also mirror the changing face of the division of social class in the U.K. In 1950, 65 per cent of individuals were employed in manual jobs compared to 47 per cent in 1991 (Financial Times 1992). One example cited is that over this period those employed in manufacturing or mining fell from 29 per cent to 23 per cent.

Of the 7 per cent of women aged 16 to 20 years 61 per cent belonged to the lower soci-economic groups, or were unemployed (table 6.2). In addition 88 per cent of them were unmarried (table 6.3). One in 3 pregnancies occurring in the under 20 years age group is terminated. However, where abortions are difficult to obtain, middle class girls are more likely to have abortions than those from working class families who tend to have their babies (Scottish Home and Health Department 1991). This too may be reflected in the

social class break down of the younger age group in this study. A further consideration is that the high number of unemployed within the younger age group (32%) could relate to their not having had time to secure employment.

The concern with this age group is that within this study they can be identified as potentially disadvantaged in relation to prenatal CF carrier screening. Firstly, almost 40 per cent had not previously heard of CF (figure 6.2) and in addition, were significantly more likely to find pre-screening information difficult to understand (figure 6.3). Fifty per cent of their partners had not read the leaflet (figure 6.4) which may reflect the fact that only 11 per cent of 16 to 20 year old women were married (table 6.3). Despite single women being significantly less likely to discuss the test with their partner (51%) than those who were married (75%) more single women discussed the test with a significant other person, usually a relative (figure 6.5). One study found that when teenagers were asked to identify the first person to whom they would turn for questions about sex, contraception and relationships a majority cited their mother (Allen 1987). Therefore, among these young women, of whom few are married, parents may have an important contribution to make in the screening decision process.

The significance of considering not only the mother but, in addition, other family members during decision counselling has been communicated (Richards 1987). Previous studies have found only a minority of women fail to discuss prenatal screening and diagnostic tests with their partner (Rothman 1988; Elkins et al 1986). In this study a total of 31 per cent of women stated that their partner had not read the leaflet (section 6.1.2.4 page 143) and approximately the same number had not discussed the CF test with him. This may reflect an apparent male perception that the decision to undergo prenatal screening is predominantly a woman's responsibility (Sjogren 1992). Most married

women stated that their partner had read the leaflet (73%) and that they had discussed the test with him (75%). A 36 year old para 1+1 offered evidence of this when she wrote:

"On reading the leaflet my immediate reaction was that I felt I would find coping with a baby with CF very difficult. Having discussed the issue with my husband we are agreed that I should have this test."

A test which may necessitate the male partner's involvement could cause a dilemma for women whose relationship with their partner is unstable. An 18 year old single primigravida told the researcher that she had discussed the test with her partner and at greater length with her mother and wrote:

"I would like to have this test but the father of the baby does not want to have it done - I am very sorry."

Fifteen per cent of married women and over 20 per cent of single women claimed not to have discussed the test with anyone other than the midwife at the booking clinic (figure 6.5). Brown (1986) revealed that male partner's sharing an interest in the pregnancy was considered by women to be the most important indicator of their partners support during pregnancy. Only 50 (2%) women claimed to have discussed the test with their general practitioner. This may simply reflect a lack of opportunity to do so. Although general practitioners were notified about the screening trial by an announcement in the local GP newsletter, when a woman first visited her GP early in pregnancy she would not have received the pre-screening leaflet and in most cases would be unaware of the test. Few women consult with their GP between having their pregnancy confirmed and attending the booking clinic. A mere 9 (0·4%) women had consulted their health visitor about the test, yet 50 per cent of women in the study already had a child and would have been expected to be in contact with a health visitor. Guilbert and Cheater (1990) found that health visitors felt their knowledge of genetics was inadequate. It is possible to

speculate that women were not encouraged to discuss genetic screening or, it may simply reflect women's perceptions of health visitors as only interested in the child (Briscoe 1989). Further research is needed to explain why so few women either felt the need or deemed it inappropriate to discuss the test with a health care professional outwith the antenatal clinic.

Most women (60%) were aware of their carrier risk of 1 in 25. What was not clear was how women perceived the degree of this risk. One study showed that the distinction between actual risk and perceived risk was an important factor in the decision to undergo prenatal diagnosis for Down's syndrome (Marteau et al 1991a). How individuals interpret genetic risks has been explored by Pearn (1973). The conclusion drawn was that individuals firstly form a subjective personal view of the disease in question and then form a subjective interpretation of the risk for that concept of the disease. A study involving 1,000 women who had accepted the offer of prenatal CF carrier screening asked them to state how they visualised a 1 in 25 risk of being a CF carrier: 84 (8%) women perceived this to be a high risk; 126 (13%) thought it was a medium risk; 337 (34%) viewed the risk as low; and 453 (45%) stated they had no perception of the risk (Mennie et al 1992 unpublished data).

No correlation was found between a woman's perception of her risk of being a carrier and her emotional response to the screening test. This might have been anticipated among women who stated that their carrier risk was as high as 1 in 4 (Table 6.4). One woman wrote:

"I feel anxious in terms of what the result will be but reassured to have as much information prior to the birth as possible."

Seventy one per cent of 16 to 20 year old women and 52 per cent of 21 to 25 year old women were unaware of their carrier risk (table 6.4) This is a concern which when coupled with the finding that 40 per cent had no prior knowledge of the existence of the disease for which they were being screened (figure 6.2) and that 50 per cent of their partners had not read the leaflet (figure 6.4) nor entered into discussion about the test supports the need to explore how this group can best become informed before they are screened. The concept of full genetic counselling for these women seems justified, however, the draw back is that it is expensive, could cause psychological morbidity in large numbers of women who have no need to be concerned because they are not CF carriers, and would be difficult to incorporate into a busy antenatal clinic. A sobering conclusion is that these women may experience corresponding difficulty with all other prenatal screening test information leaflets and scant knowledge on matters relating to pregnancy. Further research is required to explore a variety of approaches to delivering pre-screening information to the younger mother. There is evidence that young women retain information if presented in narrative form because they identify with the characters in the stories (Crow et al 1972). In relation to general health information the poorly informed report the use of television while the better informed report the use of print media such as newspapers, books and magazines (Gombeski 1981).

A considerable majority of women (85%) had previously heard of CF, and claimed to find the leaflet easy to understand (94%). More stated that they felt reassured (43%) rather than anxious (25%) about being screened. However, this leaves 32 per cent of women who seem not to know how they feel toward being screened. More detailed research might reveal more precisely how this group feel and among those who feel anxious, what exactly they feel anxious about. The results of a study examining the reasons why women accepted or declined the CF carrier test are presented in the following section.

6.2 Question two: What factors influence a woman to accept or decline prenatal CF carrier screening?

6.2.1 The sample and method of collecting data

A pre-screening leaflet inviting women to be screened outlined the purpose of prenatal CF carrier screening and explained the testing procedure. The leaflet emphasised that a negative test result would not guarantee an unaffected child but would greatly reduce the risk. It further emphasised that most couples who had a child with CF had no family history of the disease. The leaflet also stated that if a woman did not know who the father of her baby was, then it was inadvisable to take the test. Prenatal diagnosis was discussed as one option for couples where both partners were CF carriers and termination of pregnancy cited as one option if the prenatal diagnostic test showed the baby had CF. At this adjunct it was stressed that prenatal diagnosis was not recommended for couples who found termination of pregnancy unacceptable.

A self-administered questionnaire was enclosed with the CF leaflet. Question nine asked if a woman: a) had decided to have the test; b) had decided not to have the test; c) had not decided. It further asked if a woman would write in a few of her own words why she had made this decision or remained undecided. Quotations were then transcribed verbatim to a computer database and examined word by word to abstract meanings or themes which were categorised and then coded according to Field and Morse (1990).

2,207 women who were eligible for CF carrier screening were invited to participate in this study of whom 2,058 (93%) returned a questionnaire. Of these women, 1728 (84%) decided to be screened and 1362 (79%) commented on their decision. 214 (10%) had made the decision to decline the test and all commented on their decision. A group of 116 (6%) women were undecided about screening. Of these 116 women 70 (60%) were

subsequently screened and 46 (40%) declined. All were counselled by the genetic nurse and all commented on why they felt undecided. Reasons why women felt undecided were of special interest since they might depend upon the midwife to assist them resolve their uncertainty.

Themes were recorded in order of apparent emphasis which were, in the majority of cases easily identifiable. The considerable number of comments enabled a quantitative analysis of themes to be carried out.

6.2.2 Presentation of data

6.2.2.1 Characteristics of the study population

Data was available on the characteristics of all 2,058 women who returned a questionnaire of whom 1798 were ultimately screened and 260 women declined. These are summarised in (Table 6.5). Significance of characteristics was evaluated by the Chi squared test.

Analysis of subjects' parity showed that multiparous women (68%) were significantly more likely to decline screening than primiparous women (60%) (X^2, $p < 0{\cdot}05$) Women who declined the test were significantly more likely to decline MSAFP screening (X^2, $p < 0{\cdot}001$). Women who declined screening were significantly more likely to feel anxious about being screened (X^2, $p < 0{\cdot}01$). There were no other significant differences between those women who accepted screening and those who declined.

Table 6.5 Characteristics of the study population n = 2058
(numbers and percentages)

	accepted screening n =1798 (87%)	declined screening n = 260 (13%)
Age (years)		
mean	28·7 ± 5·56	27·74 ± 5·14
range	16-44	16-41
Parity		
0 + 0	720 (40%)	85 (33%)
0 + 1 >	196 (11%)	30 (11%)
1 + 0 >	882 (49%)	145 (56%)
Gestation (weeks)		
mean	12·25 ± 2·12	12·72 ± 3·63
range	6-18	7-18
Marital status		
married	1316 (73%)	199 (76%)
single	409 (23%)	49 (19%)
divorced	46 (3%)	7 (3%)
separated	24 (1%)	5 (2%)
widowed	3 (-)	0 (-)
Social class		
1	227 (13%)	35 (13%)
2	553 (31%)	80 (31%)
3	601 (33%)	73 (28%)
4	158 (9%)	25 (10%)
5	91 (5%)	14 (5%)
unemployed	141 (8%)	22 (9%)
student	27 (1%)	11 (4%)
Religion		
Protestant	997 (55%)	130(50%)
Roman Catholic	243 (14%)	50 (19%)
Christian	47 (3%)	13 (5%)
Other	42 (2%)	15 (6%)
None	469 (26%)	52 (20%)
MSAFP screening		
accepted	1776 (99%)	142 (55%)
declined	22 (1%)	118 (45%)
***Emotional response to screening**		
Anxious	422 (23%)	99 (38%)
Reassured	880 (49%)	12 (5%)
Don't know	494 (28%)	146 (57%)
* 5 no response		

6.2.2.2 Reasons why women accepted CF carrier screening.

From the comments of 1,362 women who had already decided at booking to accept screening, 7 themes emerged which influenced their decision to accept the offer of prenatal CF carrier screening (table 6.6).

Table 6.6 Themes of women who accepted CF carrier screening

Themes	Number of women
1. Early diagnosis	842 (62%)
a) prevention affected child	353 (42%)
b) knowledge about the baby	202 (24%)
c) to make an informed decision	127 (15%)
d) preparation for an affected child	87 (10%)
e) concern for the baby	49 (6%)
f) personal factors	24 (3%)
2. Reassurance	220 (16%)
3. Knowledge of carrier status	123 (9%)
4. Logic	70 (5%)
5. Simplicity of test	48 (4%)
6. Availability of test	34 (2%)
7. Precaution	25 (2%)
Total	1362 (100%)

1a) Early diagnosis

The potential to diagnose CF in the fetus was the primary reason given by a majority of women who were screened. A total of 842 (62%) women stated that they wished to

know if their baby was at risk of having, or was affected by the disease. Within this group varying reasons were given for wishing to acquire this information.

For 353 (42%) of 842 women who cited prenatal diagnosis as their reason for wishing to be screened, the intention was to prevent the birth of an affected child or to have a healthy baby. Evidence of this is apparent in the following statement:

" To spare the child and ourselves a difficult life if positive."

Some were quite blunt:

"Because we don't want a baby with CF genes."

Others took a moral stand:

"I feel that as a parent it is my responsibility to try and conceive a healthy child. Bringing a child into the world with the knowledge that he or she has a serious handicap and life threatening condition is selfish, affecting all family members and the child."

Within this group, who wished to prevent the birth of an affected child, were 65 (18%) women who revealed that they had personal experience of CF. One expressed her reason for wishing to be screened as follows:

"A friend died at 15 with CF. I wouldn't wish this on any child."

And another wrote:

"A close friend has this illness and I have seen how this has affected him, and I would not like my own child to suffer like this."

A couple where both partners were doctors revealed the effect of their experience of the disease:

"We have both seen and treated children with cystic fibrosis - we would wish to terminate our pregnancy should the foetus be affected."

One young mother recalled a recent traumatic experience which influenced her screening decision:

"I have already had my youngest son tested for CF. He was clear but while waiting for the result I found out a lot about the disease. I could not bring a child into the world to suffer and die prematurely."

Some wished to prevent the birth of an affected child on the grounds that they would be unable to cope. This view was more prevalent among primiparous women. As one woman stated:

"I do not think I could cope with a child with a major illness."

Others wanted a healthy baby:

"I basically want a healthy baby and welcome the opportunity to be screened at an early foetal date."

One woman indicated that she regarded the test as crucial in her pursuit of a healthy baby:

" Will do everything to ensure my baby is healthy."

Another, wishing to achieve the same objective wrote:

"We want our child to be born fit and healthy and have a chance in life without anything like CF holding (him - her) back."

1b) Knowledge about the baby

A total of 202 (24%) of women were influenced by the possibility of prenatal diagnosis because they wanted to know before birth if their baby had CF.

One woman wrote:

"If there is anything wrong with the baby we would like to know in advance."

and another similarly outlined her view :

"It will be good to know if my baby, before it is born, has any chance at all of having the disease. My husband agrees with this as he is anxious to know of any abnormalities."

Women in this category unlike those in others, did not reveal what action they would take as a result of gaining this information. For example:

"I'd like to know if our baby could be that 1 in 4 to have it."

Another woman similarly stated:

"To have as much information as is available about our baby's health in the early stages of pregnancy."

1c) To make an informed decision

To enable an informed decision was the reason 127 (15%) women cited early diagnosis as influencing their decision to be screened. One woman wrote:

"If, at this stage of pregnancy, I discovered there was a good chance of having a child with cystic fibrosis then I would still have time to consider another option or come to terms with it. It all comes down to having a choice."

Another echoed the same sentiment:

"I would like to know any conditions my baby has to allow time to decide whether to terminate or be able to find out more about the condition and how to deal with the problems associated before the birth of the baby."

1d) Preparation

Preparation was a motivating factor among 87 (10%) women in this group who referred to early diagnosis as their reason for wishing to be screened. Approximately 10% of them wished to be wholly prepared, typically commenting as follows:

" To be prepared for any eventuality."

A number articulated their plan of campaign:

"We decided to have this test so if we are found to be carriers we could prepare ourselves as much as possible, find out more information and advice."

Preparing themselves for the birth of a child who suffered from CF was at the forefront of the minds of a majority within this category who wished to be prepared. The following statement is representative of this groups' comments:

"If our child was to be CF we would like to be prepared."

Another echoed the thrust of this comment:

"I'd want to know if there was anything wrong with my baby, so that I can prepare and learn how to cope with this illness and try to overcome the problems that arise from it."

Others felt that they had a right to know if their child was affected:

"If it is possible to find out if your child is going to be born physically disabled, then I think we have the right to know beforehand, so that we can at least be slightly prepared for what is to come."

A similar view was repeated in the following statement:

"I think my partner and I have a right to know if we are carriers of the CF gene and so be prepared for the possibility of having a child with cystic fibrosis."

1e) Concern for the baby

For 49 (6%) of women who wished to avail themselves of early diagnosis through screening, concern for the baby was uppermost in their minds:

"Mainly concern on the baby's behalf - should find out as soon as possible."

Others revealed more deeply their feelings of concern for the baby:

"If I can prevent any suffering for my baby I will. It is wise to take the test as a precaution but termination is still a difficult decision."

A number felt responsibility toward their child's health:

"So that my husband and I feel reassured that we are doing everything possible to safeguard our baby's health."

1f) Personal reasons

Twenty four (3%) women stated that they had personal reasons for wishing to know if their baby had CF. Some already had a child with special needs.

"Well, my first baby has cerebral palsy and I don't want anything to happen to this baby."

And another wrote:

"I have already lost a baby with osteogenesis imperfecta."

Another voiced concern that:

"The father of my child is my cousin so I thought the test would be a good idea."

2 Reassurance

A total of 220 (16%) of the 1,362 screened women respondents sought reassurance from the screening test.

"I have decide to have the test done to reassure us a bit."

Although the limitations of the test were acknowledged it did not discourage this group of women from being tested:

"Reassurance, although I know the test is not 100% guaranteed."

Clearly the offer of the test increased awareness, which in turn was a motivating factor:

"Now that I am aware of the statistics concerning carriers, I would feel happier to know that I am not."

Some expressed overt concern:

"I have decided to take the test to hopefully feel some reassurance at the end result - although I do not know what I would do if the test proved positive -this makes me extremely anxious."

Another echoed this feeling:

"I would like the test just to reassure me and my husband a little. But I will worry a lot until I get the results."

Reassurance specifically about the health of the baby was being sought by some, as the following statements indicate:

"Any test that can help you feel reassured that your unborn child is healthy is worth having."

Another woman wrote:

"We are both anxious of the progress of the pregnancy and want to be reassured that all is going well."

Others sought reassurance about themselves:

"Just to make sure if I am okay!"

and:

"Because I don't think I have it but would like to put my mind at ease."

One woman appeared to be relating symptoms she experienced to those of CF and wrote:

"I have a lot of belly pain."

The expression *"peace of mind"* was used by 50 per cent of women who sought reassurance. Alternative statements were: *"to put my mind at ease"* or *"to make my mind rest."* Many women used these expressions in conjunction with other statements:

> *"To find out if I am a carrier and to give me peace of mind that I will hopefully have a healthy child."*

A further examples illuminates the need for reassurance:

> *"Cystic fibrosis is a very serious disease, would have peace of mind if the test proved negative."*

3. Knowledge of carrier status.

The study revealed that 123 (9%) of those women who intended to be screened, wished to know if they were a CF carrier:

> *"I think it is important to know if you or your partner have the CF genes."*

Some indicated their reason for wishing to know if they were a carrier:

> *"I would like to know if I was a carrier and if I am I would like to know my husband's carrier status so that we can be as aware as possible of potential problems."*

Others wished to know if they were carriers but indicated that the information would not affect the outcome of the pregnancy:

> *"I would like to know if I am a CF carrier. This would not change my decision upon having the baby."*

4. Logic

In total 70 (5%) women gave reasons of logic for accepting the screening test, for example: *"It appears logical"* or *"felt it was the sensible thing to do"* or *"there seems no reason not to do so."*

Aside from a few, this group of women offered terse statements containing little sign of emotion. A rare exception to this commented thus:

"I think it's sensible to have the CF test seeing as other tests are carried out at the same time. It helps to reassure you that every precaution is taken."

5. Simplicity or non-invasiveness of the test

The non-invasive nature of the mouth wash test was an important factor in the decision of 48 (4%) of women to be screened. Comments representative of these women were:

"The test is simple and reasonably reliable and there is no reason not to have the test considering the consequences should the child be affected with the CF genes."

or:

"Because it is a simple mouth wash test which has no side effects on the baby."

Another wrote:

"I have no reason not to take the test. My GP says it won't harm me or the baby. If anything was wrong I'd rather know about it."

6. Availability of the test

Thirty four (2%) women stated that they were being screened simply because the test was available. A few appeared indifferent in their attitude toward the test:

"I am having the test simply because it is being offered to me."

Others indicated they felt they did not wish to refuse something which was offered:

"I think it would be better to have the test and know you are not missing out on anything and it's very simple."

Perhaps some felt the decision had already been made for them by offering the test:

"Because the test is available and I feel it must be important if you are bringing it to our notice."

A majority of women who took the test because it was available viewed it enthusiastically:

"Testing of this nature is a wonderful advance and should be taken advantage of"

However, some were more discriminating:

"If the test is available I may as well have it, if no risks are carried to myself or unborn child."

7. Precaution

Twenty five (2%) women stated that they were accepting the test as a precautionary measure but none gave any hint as to their likely action should the test prove positive. Comments written were comparable to the following:

"To be on the safe side" or *"Better to be safe than sorry."*

6.2.2.3 Reasons why women declined the test.

Of the 279 (13%) women who had made the decision to decline screening , 214 (77%) commented on why they had declined the test. There were nine categories which emerged. These are listed in table 6.7.

Table 6.7 Themes of women who declined CF carrier screening

Theme	Number who declined (numbers and percentages)
1 against termination of pregnancy	124 (58%)
in general	98 (79%)
specifically for CF	26 (21%)
2 for reasons of male partner	17 (8%)
3 considered risk of a CF child low	15 (7%)
4 error rate of test unacceptable	14 (7%)
5 test would cause anxiety or worry	11 (5%)
6 don't want test during pregnancy	11 (5%)
7 don't want to know	9 (4%)
8 too difficult a decision if test positive	8 (4%)
9 no reason	5 (2%)
total	214 (100%)

1. Against termination of pregnancy.

The main reason why respondents declined screening was opposition to termination of pregnancy. A total of 124 (58%) women who had made the decision to decline the test cited this reason. Most (79%) of this group were against termination in general. As one woman wrote:

> *"For us killing our unborn child because of any disability is not an acceptable option. I know of no other reason why the test would be helpful, so I wouldn't have it."*

Against termination of pregnancy for CF

A further 26 (21%) women opposed to termination stated that they held this opinion specifically with regard to the disorder CF. The following comment typifies statements made by this group:

"I don't think in these circumstances I would find termination acceptable, so I'd prefer not to start on this route."

2. For reasons of partner

The CF carrier test is distinct from most other prenatal screening tests with respect to involving the male partner. Indeed prior indication that a woman's partner would be willing to be screened if indicated, was paramount.

Seventeen (8 per cent) of those who declined screening did so because their partner had indicated his reluctance to participate. More than one woman commented along the following lines:

"My husband would be far too busy to have the test done if it became necessary"

Another explained:

"It's my partner - he doesn't really know about the pregnancy and he says he hasn't got anything like that - he says it will take some time for him to think about it."

3 Considered risk of having a child with CF was low.

Perceiving one's risk of having an affected child as low clearly influenced women to decline the test. A total of 15 (7%) respondents who planned to decline the test stated this was their reason for refusing. Of these, 14 (93%) were multiparous women. A typical statement was as follows:

"As I already have a completely healthy child I don't think I am at high risk of having a child with CF. Also there is no family history."

4 Error rate of the test unacceptable.

The inability of the CF carrier test to detect all carriers was a factor which influenced 14 (7 %) to refuse the invitation to be screened. This reason frequently overlapped with their concern that an inconclusive test result would generate anxiety throughout the remainder of the pregnancy. One woman summed up the general view of this group:

"As only 85% of cases tested are successful, if my test is negative I'm going to be anxious that I might be that 15% and as you state you can't guarantee the child will not have CF."

5 Test caused anxiety or worry

Eleven (5 %) women stated that the test would generate unacceptable levels of anxiety and for this reason they would prefer not to be screened. One woman envisaged the following scenario:

"The worry of finding out I have a single gene until my partner is tested. If the test is negative the worry is unnecessary. If he is positive, even more worry would result until the prenatal diagnosis when if the baby is negative, again the worry has been unnecessary."

6 Don't want test during pregnancy.

Eleven (5 %) respondents who declined the test stated they did not wish to be tested during pregnancy. Six (55%) women indicated that they would have accepted screening either before pregnancy, or in the postnatal period as part of future reproductive decision making. One subject explained:

"I feel it is now too late for the test to be relevant. If the test was available for someone intending to become pregnant but not yet pregnant I would have had it at that stage."

7 Don't want to know

Nine (4 %) women simply said *"don't want to know"* but gave no reason for their decision.

8 Too difficult a decision if test positive

The thought of being screened in pregnancy concerned a group of 8 (4 %) respondents because of the prospect of having to decide among options raised by a positive test result. One woman reasoned:

> *"I think I would face a very difficult moral dilemma if I discovered whilst pregnant, that both my husband and I were CF carriers. I would then want to have the baby screened and if it had CF I would be very worried about making a decision to have an abortion, which in theory I'm opposed to, but realistically, don't know what I'd do."*

Of this group, six (75%) had either a history of infertility or previous pregnancy loss which they felt contributed to their apprehension which revolved around the prospect of having to confront a dilemma about continuing a much wanted pregnancy. One woman wrote poignantly:

> *"My husband and I have decided against the test because we have waited a while for this baby. To find out something was wrong would be shattering for us both, we would rather take our chances and hope everything will be okay."*

9. No reason.

Five (2 %) women stated they had no reason for refusing claiming they simply did not wish to be screened. One lady stated:

> *" Don't know - just don't want to have test done."*

6.2.2.4 Reasons why women were undecided about screening

A total of 116 (5%) women who were initially undecided about being screened could be divided into 12 categories (Table 6.8). These were considered an important group who might require additional information or counselling from the midwife.

Table 6.8. Themes of women who were undecided about CF carrier screening

Theme	Accepted	Declined	Total
1 required more information	31	10	41 (35%)
2 for reasons of male partner	9	9	18 (16%)
3 against termination of pregnancy		17	17 (15%)
in general		14	
specifically for CF		3	
4 considered risk of CF child low	7	4	11 (10%)
5 test caused anxiety or worry	6	2	8 (7%)
6 too difficult decision if test positive	4	2	6 (5%)
7 don't understand the test	4		4 (3%)
8 not screened in previous pregnancy	4		4 (3%)
9 just undecided	4		4 (3%)
10 error rate of test		1	1 (1%)
11 test might harm baby	1		1 (1%)
12 advances in treatment for CF		1	1 (1%)
total	70	46	116 (100%)

1 Required more information.

Of the 116 (5%) women who were undecided about CF carrier screening 41 (35 %) wished more information about the implications of screening.

One woman stated:

"Want to know more about the tests that are carried out before making a decision."

Another echoed the same thrust:

"Because I would like more information on the subject if possible as the leaflet does not explain enough."

Thirty one (76%) of those who required more information accepted the test and 10 (24%) declined.

2 For reasons of partner.

Eighteen (16%) were uncertain for reasons of their partner. One woman summed up her situation when she wrote:

"Partner would rather not know. I'd like to be prepared for it if it happened."

And another wrote:

"I want to make sure all is well - to be able to make a well informed decision if the need arises. I want the best for my baby. My husband is away and won't be returning for a few weeks."

Of this group half accepted and half declined the test.

3 Against termination of pregnancy.

Seventeen (15%) respondents stated that they were undecided about being screened because they were either against or doubtful about termination of pregnancy. Most 14 (82%) were against abortion for any abnormality. Three (18%) women were doubtful about termination specifically for CF. The following statements describe how many in this group felt:

"Want to take the test for reassurance if negative but, if its positive I doubt if I'd want an abortion so why have the test? "

Another commented:

"Although I can see the benefits in such a test I feel for me it would be wrong as I would not have an abortion even if my child had CF and it seems pointless to know before the birth."

All the women in this group ultimately decided against being screened.

4 Consider the risk of a CF child low.

Eleven women (9%), among those who were undecided, felt they were probably at low risk of having an affected child. A number had not understood that a family history of CF was not a prerequisite to being a carrier. As one woman wrote:

"Well none of my family have ever had it and I don't think the father of the child or his family have got it."

Others, since they already had unaffected children, wanted to know how this affected their risk:

"Having had 2 children already who are perfectly healthy, I am going to check with the doctor, if there's more of a chance or less of a chance."

Of these 11 women 7 (64%) were screened and 4 (36%) declined.

5 Test causes anxiety or worry.

Eight (7%) women felt uncertain about being screened because of generating anxiety.

"Whilst we would rather know in advance if our unborn child has CF, the possibility of 3 periods of stress / anxiety awaiting test results doesn't appear conducive to achieving the state of mind desirable during pregnancy - particularly as the chances of both partners being carriers is low."

This lady was among six (75%) women in this group who decided to be screened and a further two (25%) women declined.

6 Too difficult a decision if the test was positive.

Six (5%) women were undecided about the test because of the anticipated dilemma they might face if the test proved positive. One woman explained her uncertainty stemmed from:

"Being worried about the decisions to be made as a result of the knowledge it may give. Still undecided as to the best thing to do, consequences for child affected etc."

Of this group 4 (67%) women decided in favour of being screened and 2 (33%) women declined the test.

7 Don't understand test.

Four (3 %) women expressed their lack of understanding of the test. Two examples of their statements help describe their difficulty:

" I don't know if I want the test because I don't really understand the treatment. In one part it says its just a mouth wash and in the next it goes on about a screening test and I don't understand it. In fact I think it's quite frightening."

And another wrote:

"Because me and my partner don't understand the carrier testing. We would change out desin (decision) if you could tell us more about it." (sic)

All 4 subjects in this group ultimately made the decision to be screened.

8. Not screened in previous pregnancy.

Four women (3%) stated that they were ambivalent about being tested because the test had not been available at the time of their previous pregnancies. All subsequently decided in favour of being screened. Their view is described in the following two statements:

" Happy with my last two pregnancies and will just have the usual tests."

and:

" Because this did nor exist when in my other 3 pregnancies." (sic)

9. Undecided or Don't know what to do

Four (3%) women were undecided and confused about what to do for the best. One young woman explained her dilemma:

"I don't know, because there are, or seem to be, so many diseases or illnesses that could affect unborn babies - I find it all very worrying."

and another said touchingly:

"I just want the best for my baby."

All four women decided to be screened.

10 Error rate of test unacceptable.

The error rate proved to be unacceptable to one (1%) woman who ultimately decided against being screened:

"The test is not 100% successful. If it can't guarantee diagnosing the possibility of having a child with CF then I would not feel reassured to have a negative result even if the risk becomes less."

11 Because the test might harm the baby.

One (1%) woman who decided in favour of being screened was initially uncertain because of fears that the test might harm the baby commented:

"I feel I would like to know more about the test e.g. is it simply a mouthwash test or are there any follow-up tests further on into the pregnancy which might harm the baby. I feel more disposed to having the test than not."

12. Because of advances in treatment or research of CF.

Finally, one (1%) woman who was undecided declined the test and did so because of advances in treatment for CF and ongoing research. She wrote as follows:

"I would not terminate a pregnancy if CF was diagnosed. Having worked with cystic children and adolescents, medical advances are being made which increase both the quality of life and the life expectancy of the CF sufferer."

6.2.3 Discussion

A total of 1692 (77%) of the 2207 women eligible for screening, commented in writing on their decision about the CF carrier test. Their comments gave an insight into their own individual perceptions of the test and its appropriateness based on their own value systems. The study revealed the many and varied factors which women stated were important in their decision to accept or decline CF carrier screening during pregnancy.

These ranged from societal to individual. It also confirmed that women will voice their opinion and feelings about new developments in technology when given the opportunity.

The invitation to pregnant women to write about their wishes and concerns has been carried out in a previous study and found to be beneficial both to a woman and to the health care professionals concerned (Jones 1990). During her study Jones examined the antenatal records of 50 women to identify birth plan requests. In 45 cases the records stated 'no specific requests'. Then, when subsequently asked to write their wishes women did so in a clear and detailed account. Educational background and career attainment did not affect the response rate.

Technological developments challenge women to confront issues which involve questioning their feelings about motherhood (Rothman 1988). They are compelled to confront their feelings toward the fetus and the normal process of maternal-fetal bonding. But the fetus is not their sole consideration, women feel additional responsibilities to other family members and are aware of their own coping ability. Most women appeared to have had little difficulty in deciding whether not to be screened with only 5 per cent of women intimating they had difficulty reaching a decision.

The factors influencing women in their decision were sometimes overlapping and interlocking. Initially five fields in each database record were allocated to theme categories but this was quickly reduced to three. Themes were recorded in order of apparent emphasis which were, in a majority of cases, easily identifiable.

Kitzinger (1987) expresses the view that many women feel guilt that they may be seen to cause delay in the clinic process by questioning or firmly pursuing what they want. The invitation to respond in writing may have advantages over an inducement to

respond verbally. First, it may be regarded as a more candid invitation, secondly it allows the individual time to assimilate their thoughts, and thirdly, and perhaps most importantly, it may encourage the individual to realise the implications of their decision and hopefully avoid accepting a test simply because they feel a need to conform.

The reason most women (62%) accepted the test was for reasons of early diagnosis of CF in the fetus. There was, however, a range of motives cited for wishing to know if the baby was affected or at risk of having CF. These ranged from prevention of, to preparation for the birth of an affected child. Although previous studies have indicated that a majority of women accept prenatal screening to receive reassurance (Farrant 1985; Davies and Doran 1981) only 16 per cent of women indicated that this was their sole reason for accepting CF carrier screening.

The comments made suggest that women are discriminating in their reasons for accepting prenatal tests. This is an encouraging finding because concern has been expressed that women feel unable to exercise the options which prenatal screening and diagnosis offered. This is blamed on a lack of information and emotional support which reduces women's autonomy and ability to make free and informed choices (Farrant 1985). One reason for this variation could be that the media have played a significant role in educating and enlightening women who may now recognise and apply their consumer power.

A study of health related topics in six of the most widely read women's magazines in the USA over a two year period found an average of three health related articles in each of the magazines studied. There was an emphasis on health promotion and protection, family and personal health practices. The best informed groups were women and young adults (Miller et al 1981). Readers of these journals were found to be of the higher social

classes and well educated. In the UK, newspaper articles about genetic screening for CF carriers have appeared mainly among broad sheet newspapers in their science and health columns (Wilkie 1992; Carrell 1992; Christie 1992; Cookson 1992).

Television has introduced genetic conditions including CF through documentary programmes and popular drama programmes. Delivery of information particularly through drama programmes or true stories may play a crucial part in individual retention of information, particularly if viewers or readers identify with the characters portrayed (Crow et al 1972). An article in the woman's journal "Family Circle" told the story of how the actress Jenny Agutter discovered during her first pregnancy that she was a CF carrier. The article gave a detailed description of the disease and the carrier screening test and technical details were illustrated by colourful diagrams. A reassuring photograph of the actress and her healthy son completed a presentation which women could easily relate to (Family Circle 1992).

Six per cent of women were explicit about their wish to prepare for the birth of an affected child; a further 127 (9 %) women wished to be able to make an informed decision; and 202 (15 %) women wanted information about their baby (table 6.6). It may be reasonable to assume a number of women in the latter two categories would likewise use diagnostic information to prepare for the birth of an affected child. Conversely, there were women who explicitly stated their intention was to terminate an affected pregnancy.

Few women (2 %) stated they wished to be screened simply because the test was available. Obtaining information about the baby early in pregnancy without having to decide immediately on a particular strategy was favoured by some. As one woman commented:

"It's a non-invasive test and there are a number of points where one can decide whether or not to proceed further."

The difficulty of anticipating women's reactions based on hypothetical situations has been acknowledged (McIntyre 1987). Nonetheless, at this early juncture 26 per cent of all women who wished to be screened stated that their intention was to prevent the birth of an affected child. Reasons cited were: their experience or knowledge of the disease; misgivings about their ability to cope with a child suffering from a chronic illness, and perceived adverse effect upon their family. As one woman explained:

"We already have 4 healthy children. I feel I have a duty to myself and family to have this test. Giving birth to a CF child would have serious consequences for the whole family - I would prefer to prevent this if I can."

The options of whether or not to proceed with an affected pregnancy were influential:

"We wish to have the test since, if we were both found to be carrying the CF gene and a prenatal diagnostic test was to show that the baby was going to have CF, we would wish to consider a termination."

The following discussion combines data from tables 6.7 and 6.8. Unquestionably, women's opinion on termination of pregnancy was the factor which motivated most to decline the test. This is a similar finding to previous studies (Davies and Doran 1981; Fadden et al 1987; Kyle et al 1988). Of the 112 women who were completely against termination of pregnancy, 78 (70%) had declined MSAFP screening. In contrast only six of the 29 (21 %) who were against termination specifically for CF declined MSAFP screening. It is interesting that 33 (30 per cent) of those who stated they were completely against termination of pregnancy, were planning to participate in the MSAFP screening programme. One reason for this may be that no reference is

made to termination of pregnancy in the Hospital's leaflet describing this test, whereas the CF screening leaflet particularly draws attention to this as a possible consequence.

The incompleteness of CF carrier testing was not a major concern. Seven per cent of women who declined the test did so for this reason. Frequently this reason was coupled with the belief that being screened would generate anxiety:

"With the test only being 85% accurate I do not feel it is worth putting myself through the worry."

A total of 13 (5%) stated they had declined the test because they anticipated it would provoke unacceptable levels of anxiety. It is interesting that so few women cited this as a reason when significantly more women who declined screening stated, in the pre-screening questionnaire, that the test made them feel anxious, when compared to women who accepted the test (figure 6.7 page 146 and table 6.6 page 156). It would appear that perhaps subconsciously women who are made to feel anxious avoid situations which may induce stress. Further research is needed to ascertain whether these women are aware of their behaviour and the reasons for their anxiety. Nine (69 %) of the 13 who refused on the grounds of anxiety did, however, accept MSAFP screening. This could be accounted for by the fact that neither the false positive or negative rates associated with MSAFP screening were intimated in the hospital's booklet outlining this test, nor was termination of pregnancy mentioned as a possible consequence. It would seem reasonable to conclude that it was the candidness of the CF information leaflet which allowed women, for whom this information created uneasiness, to make an informed choice. It is a disappointing finding that women are denied the full facts about MSAFP screening. Although they are granted the freedom to choose whether or not to accept the test, women are disadvantaged if not in full possession of the facts.

Prenatal CF carrier screening introduced a relatively new concept of screening in its involvement of the male partner. Some women were reluctant to admit the existence of a supportive partner for fear of jeopardising their entitlement to state financial benefits. However, when asked about accessibility to the male partner if screening was indicated, most claimed that their partner would be available.

Genetic conditions are frequently equated with a family history. Regardless of emphasising that a family history was not a prior condition to being a CF carrier, this was a difficult concept for some to comprehend. Multiparous women in particular were inclined to perceive that their risk of having a child with CF was low. As one subject stated:

" I have five healthy children already and feel no need for this test. "

In some instances multiparous women indicated that their satisfaction with the care given in previous pregnancies was a factor:

" Happy with my last two pregnancies and will just have the usual tests."

Only 11 (4%) women actually stated they were against being screened during pregnancy and would therefore decline the test:

"Feel it is too late to be having the test. Before pregnancy I would have wanted it, including tests for other genetic diseases."

Nevertheless, others indicated that although they planned to be screened, they would have preferred the option to have been made available prior to conceiving. A study of attitudes of recent parents to CF carrier testing found that approximately half the sample were in favour of screening in early pregnancy (Green 1992). In the present

study, because the test was offered during pregnancy, some women may have presumed that CF carrier screening was exclusively a prenatal test.

The literature search revealed that in Britain more than one in ten couples experience difficulty in either achieving a pregnancy or having a live born child (Page 1988). Moreover, studies indicate that couples who have undergone infertility investigations and subsequently encounter prenatal diagnostic procedures experience elements of the psychological trauma associated with their infertility (Sandelowski et al 1991). The CF screening test is offered at a woman's first clinic visit. The prospect of being confronted with a decision to continue or end the pregnancy was a situation which some of these couples made clear they wished to avoid.

The importance of good pre-screening information cannot be over emphasised. Studies indicate that informed decision making requires time (Lorenz et al 1985). The aim of the pre-screening leaflet in this trial was to present a global view of CF carrier screening to women and their partners well in advance of screening so that they might make a decision with which they are happy. There was no indication that women felt constrained in their decision to accept or decline the CF carrier test during pregnancy. Generally their perspective was that screening granted them a measure of control over the outcome of their pregnancy.

It has been suggested that it is doctors who wish perfect babies and not women (McIntyre 1987). This study revealed that 6 percent of women wished to be screened to prepare for the birth of an affected child. Although this is a minority group they constitute an important pre-screening counselling component. Although couples where both partners test positive could be advised that the baby was at high risk of having the disease and prepare themselves for such an event this goes against the primary objective

of prenatal screening which is to prevent the birth of infants with conditions which screening can detect. Although this objective may not be openly promoted in patient leaflets, it is currently the method of evaluating genetic screening programmes and prenatal screening programmes which are justified on the basis of the financial savings achieved by reducing the birth incidence of a number of conditions such as Downs syndrome, spina bifida, and CF (Gill et al 1987; Chapple et al 1987; Wald and Cuckle 1988; Goldstein and Philip 1990; Golbus 1992). Some recognise that there are significant non-monetary costs and benefits and that these should be considered in the organisation of such services (Clarke 1993). Indeed it is argued that economic factors should only be considered in the light of improving efficiency and only after service goals have been established (Phinn 1990). Evaluating health care services is not simply a technical, professional matter, it includes interpersonal aspects where consumer opinion is as important (Vuori 1989). The results of this study show that women have varying objectives in undergoing screening and a minority are at variance with the objectives of the providers. For a service to achieve its full potential providers need to be clear about its values and objectives, but to supply a high quality service providers need to be aware of consumer perception of its values and objectives (Vuori 1989).

Many women who accept CF carrier testing in order to promote a healthy outcome of pregnancy may be unaware of the potential stress associated with receiving a positive test result. Fortunately it is a small minority of women who ultimately have to wrestle with the moral and emotional questions of abortion. Nonetheless, there are a substantial number of women who are identified as CF carriers but whose partners will test negative. If midwives are to help them, they need to be equipped with an awareness of the possible stressful effects of being identified as a CF carrier during pregnancy. The following section presents the results of a study which assessed the psychological impact on women identified as CF carriers and their partners who received a negative test result.

6.3 Question three

Will identifying a woman as a CF carrier during pregnancy provoke a stressful response both in her and in her partner?

6.3.1. Sample and method

Of 2,207 women who were eligible for CF carrier screening, 1,812 (82%) women accepted the offer of testing and were invited to participate in this study. 1,798 (95%) agreed to participate. Among these 1,798 women were 69 (4%) women who were identified as CF carriers. In all cases their male partner was screened. Three couples were identified where both partners carried a CF mutation and they were excluded from this study. One couple suffered a pregnancy loss and one couple failed to complete the questionnaires. A further exclusion was a male partner who expressed reluctance to enter the study, however, his carrier partner wished to be included. A total of 64 carrier women and 63 male partners participated in the study.

For each carrier two control subjects of the same parity were selected. Control subjects had attended the same antenatal booking clinic as the carrier, had received a negative CF test result, and had a male partner willing to act as a control subject. A total of 116 female controls and 115 male controls were recruited. Of these 13 couples failed to complete all of the questionnaires and 2 couples suffered a pregnancy loss. A total of 101 female controls and 100 male controls participated in the study.

Socio-demographic data was obtained from carriers' and controls' antenatal records.

A self-administered pre-screening questionnaire was designed and sent to all women along with an information leaflet describing the aims of the carrier test and outlining the

screening procedure. Incorporated in the questionnaire was a 12-item General Health Questionnaire (see appendix) designed to assess the threshold emotional status of a woman prior to her being screened.

The Symptom Rating Test (SRT) (see appendix) was used to identify the nature of distress among women who presented at the clinic with a positive GHQ score.

The significance of differences in GHQ scores between groups was evaluated by the Chi-squared test. Symptom Rating Test (SRT) scores were not normally distributed (skew to higher values), therefore the significance of differences between groups was assessed by the median test.

Women were asked to complete the threshold GHQ at home and bring it with them to the booking clinic. Those who had not completed a questionnaire were asked to complete one at the clinic. Pre-screening counselling was carried out by the midwife responsible for booking the patient and a mouth-wash sample was obtained. The threshold GHQ was scored by the researcher and those women with a positive score (3 items or more) were interviewed to establish the likely reason for their response. They were asked to complete a SRT (termed threshold SRT) to determine the nature of their psychological disturbance. GHQ and SRT scores along with interview data were recorded on a computer database for ease of storage and recall when a carrier was identified.

Women identified as carriers were contacted a week later by telephone or, in a minority of cases, by letter and invited to attend the hospital, along with their partner, for genetic counselling. The couple were seen prior to genetic counselling by the researcher and the aims and sequence of the questionnaires explained. Male partners were asked to

sign a consent form and both partners were asked to complete a GHQ and a SRT (termed GHQ1 and SRT1)

Counselling was then carried out by the genetic nurse and a mouthwash sample was obtained from the partner. Couples were given a detailed information leaflet with a contact telephone number.

On receipt of the partner's negative test result (average 4 days) the genetic nurse contacted the couple in all cases by telephone and informed them of the result. A letter was sent confirming the partner's negative result and reiterating the couple's residual risk of 1 in 640 of having an affected child. Enclosed was a stamped addressed envelope and a GHQ and a SRT (termed GHQ2 and SRT2). Six weeks later the couple were sent a further postal GHQ and SRT (termed GHQ3 and SRT3) and finally six weeks after the delivery of their baby the same two measures were sent (termed GHQ4 and SRT4).

Controls were contacted by telephone in all but 4 cases where contact was made by letter. Control couples received a postal GHQ and a SRT at comparable intervals to carriers and partners.

6.3.2 Presentation of data

6.3.2. 1 Characteristics of the study population

The socio-demographic characteristics of the screened population, carriers and controls are shown in Table 6.9. There was no difference in socio-demographic characteristics between the total, carrier and control populations.

Table 6.9 Characteristics of the total screened population, carriers and controls

	total population n=1798	carriers n=64	controls n=101
age (years)			
mean	28·07	27·86	28·64
range	16-44	18-44	20-40
parity			
primiparous	916 (51%)	35 (55%)	52 (51%)
multiparous	882 (49%)	29 (45%)	49 (49%)
gestation (weeks)			
mean	12·25	11·94	12·25
range	6-18	7-16	7-18
marital status			
married	1316 (73%)	48 (74%)	82 (81%)
single	409 (23%)	14 (22%)	16 (16%)
divorced	46 (3%)	0	3 (3%)
separated	24 (1%)	1 (2%)	0
widowed	3	1 (2%)	0
Socio-economic group			
1	227 (13%)	5 (8%)	11 (11%)
2	553 (31%)	18 (28%)	31 (31%)
3	601 (33%)	23 (36%)	36 (35%)
4	158 (9%)	10 (16%)	16 (16%)
5	91 (5%)	4 (6%)	3 (3%)
unemployed	141 (8%)	4 (6%)	3 (3%)
student	27 (1%)	0 (-)	1 (1%)

6.3.2.2 Results of the General Health Questionnaire (GHQ)

6.3.2.2.1 GHQ results of the total screened population

A total of 576 (32%) of the screened population presented with a positive threshold GHQ. Of these 519 (90%) women were interviewed at the clinic to elicit reasons for their psychological distress. Failure to interview 10 per cent of women occurred either because they had left the clinic before the researcher could speak to them, or because the researcher was advised against interviewing women who were experiencing a serious complication of pregnancy. Reasons cited for presenting with a positive threshold GHQ are listed in table 6.10. Included in the total population were the 64 CF carriers who participated in this study and their 101 controls. Fourteen (22%) carriers and 25 (25%) controls presented before screening with a positive threshold GHQ (table 6.10).

Among the total screened population 38 per cent cited symptoms of pregnancy as the main cause for their psychological disturbance. Although most felt positive about their pregnancy they complained of tiredness, nausea and emotional volatility (table 6.10). Twelve per cent stated that their pregnancy was unplanned. Twenty two per cent were anxious about the pregnancy for reasons of: poor obstetric history (58 women); no specific reason (40 women); problems with the pregnancy such as threatened abortion (8 women); and awaiting the results of a chorionic villus sample or amniocentesis later in the pregnancy (10 women). Four per cent of women had a history of a previous psychiatric episode (Table 6.10).

Table 6.10 Reasons women gave for a positive threshold GHQ			
Reasons	**Bookers n = 519**	**Carriers n = 14**	**Controls n = 25**
symptoms of pregnancy	196 (38%)	5 (36%)	12 (48%)
anxious about pregnancy	116 (22%)	1 (7%)	5 (20%)
unplanned pregnancy	69 (13%)	2 (14%)	2 (8%)
active child at home	23 (4·5%)	1 (7%)	0
previous psychiatric episode	23 (4·5%)	0	1 (4%)
existing illness	12 (2%)	1 (7%)	1 (4%)
history of infertility	17 (3%)	0	0
strained relationship with partner	12 (2%)	0	1 (4%)
social or domestic problems	18 (4%)	1 (7%)	2 (8%)
unemployment	10 (2%)	0	0
bereavement	8 (2%)	3 (22%)	0
child with special needs	4 (1%)	0	0
job dissatisfaction	5 (1%)	0	1 (4%)
involuntarily apart from partner	6 (1%)	0	0

Older women (36-45 years) were significantly more likely than other age groups to present with a positive threshold GHQ (Chi-squared test p=<0.005). Younger women (16-20 years) were more likely, though not significantly, to present with a positive threshold GHQ at booking (Figure 6.9).

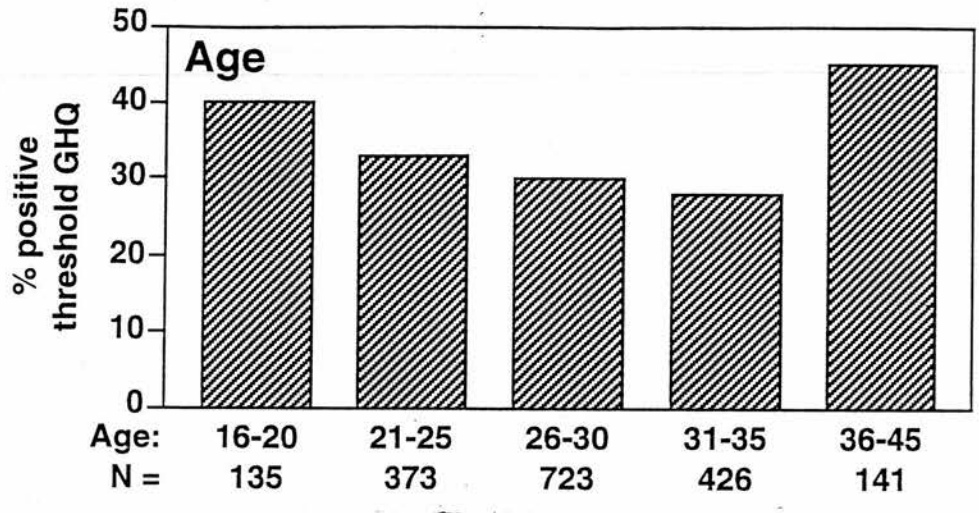


Figure 6.9 Antenatal bookers with a positive threshold GHQ; with age

There was a trend for women who were single or separated to present at booking with a positive GHQ score (Figure 6.10).

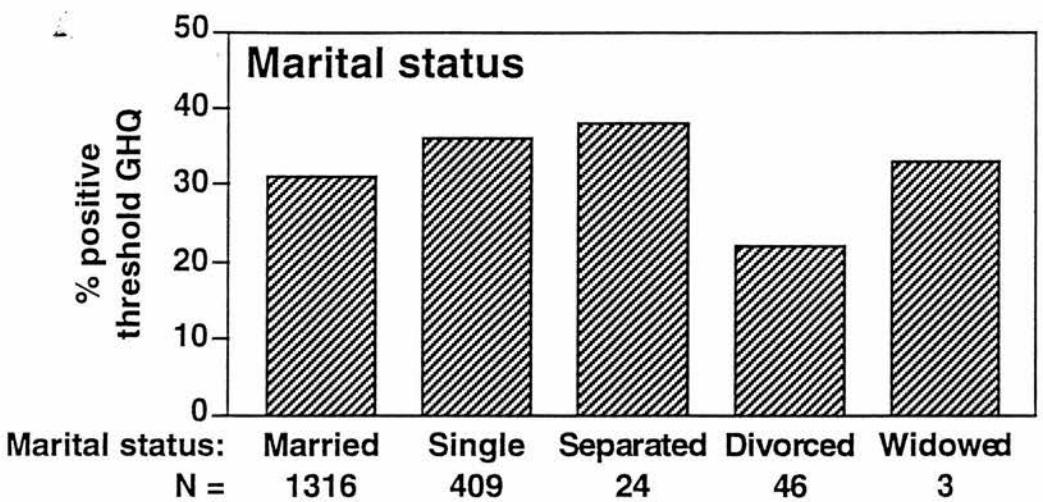


Figure 6.10 Women with positive GHQ score; with marital status

Socio-economic background did not significantly influence a woman's threshold GHQ score (Figure 6.11).

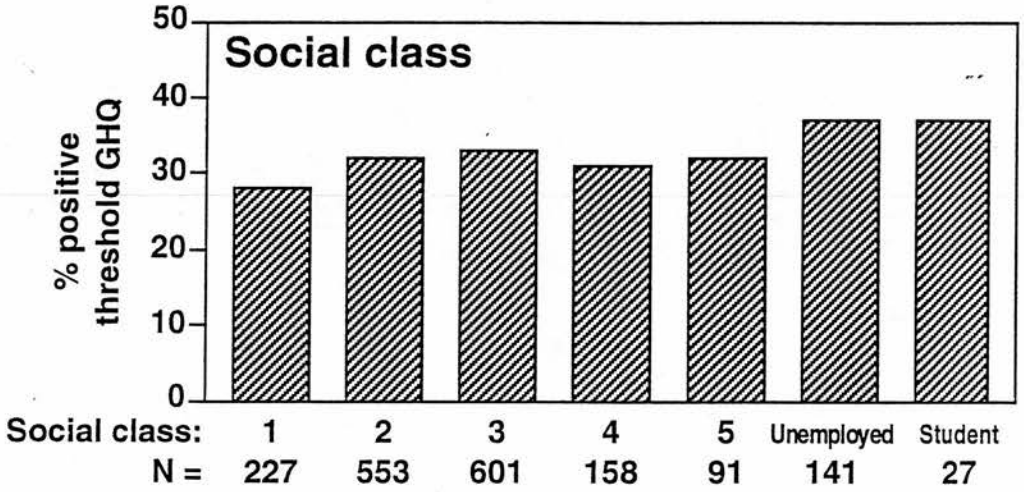


Figure 6.11 Women with a positive threshold GHQ score; with social class.

There was no correlation between a positive threshold GHQ and gestation of pregnancy (Figure 6.12).

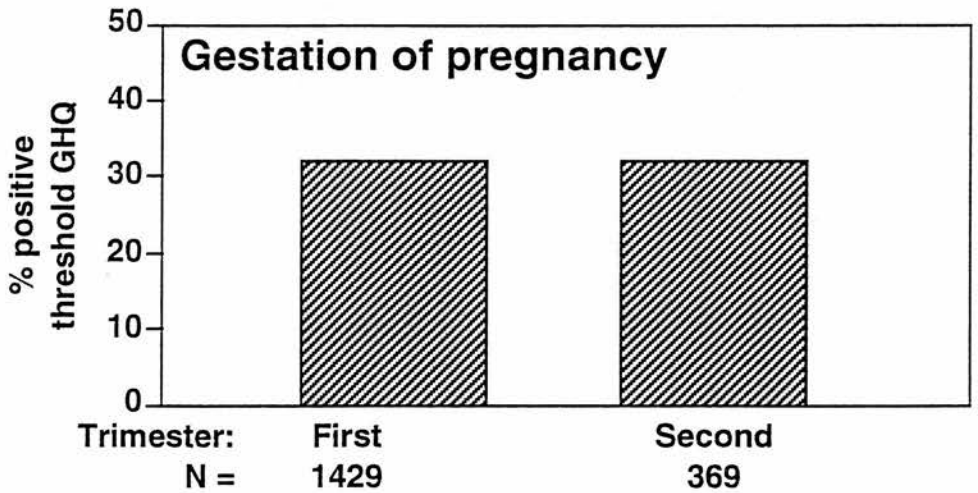


Figure 6.12 Women with a positive threshold GHQ score; with gestation of pregnancy.

Neither a woman's parity or her emotional attitude to being screened affected participants' GHQ scores at booking (data not shown).

6.3.2.2.2. GHQ results of CF carriers and controls

Sixty four carriers participated in this study along with 101 controls. The control subjects had attended the same booking clinic as the carrier but had received a negative test result. Among the 64 carriers were 14 (22%) carriers who presented with a positive threshold GHQ, compared to 25 (25%) of control subjects. The reasons these women were experiencing stress at the time of booking are listed in table 6.10.

On receiving their positive test result, the proportion of carriers (53%) with a positive GHQ1 score were significantly greater than the proportion of control subjects (27%) using the Chi-squared test ($p = <0.001$) (Figure 6.13).

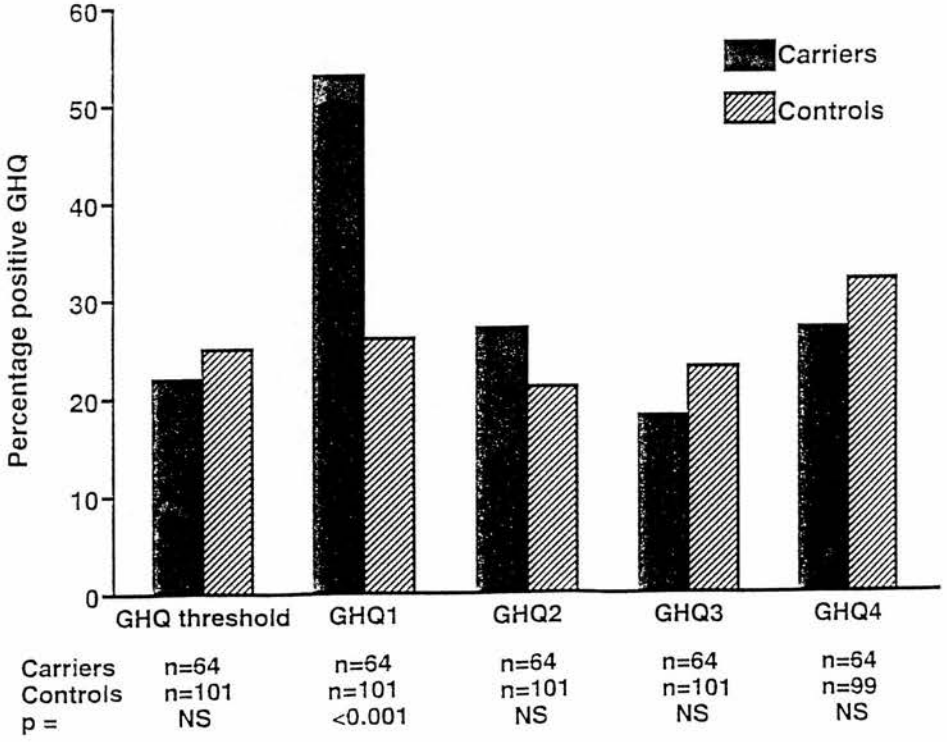


Figure 6.13 Carriers with a positive GHQ compared to controls at each of the five assessment points.

Following receipt of their partner's negative test result (GHQ2), at six weeks after the test (GHQ3) and at 6 weeks post-delivery (GHQ4), carriers showed no significant difference in the proportion of positive scores when compared to control subjects (Figure 6.13).

Five (36%) of the 14 carriers who entered the study with a positive threshold GHQ and 11 (44%) out of 25 controls maintained these scores throughout the study for reasons specified in table 6.11.

Table 6.11 Reasons why 5 carrier and 11 control subjects submitted positive GHQ scores at each of the 5 assessment points.

Carriers	Controls
First baby died previous year	Chronically sick child
Mother died recently	Hyperactive child - husband works away
Recently widowed - unplanned pregnancy	Unplanned pregnancy (3 subjects)
Husband diagnosed as diabetic	Anxious about pregnancy (2 subjects)
ECG monitoring during pregnancy for attacks of breathlessness	Amniocentesis and domestic problems
	Agoraphobia sufferer
	Moved house (2 subjects)

There was no correlation between participant's age, social class and parity and GHQ scores. However, women who perceived their carrier risk incorrectly were more likely, though not significantly so, to have a positive GHQ1 score at the time of receiving their positive test result, when compared to women who knew their risk of being a carrier.

6.3.2.2.3 GHQ results of male partners and controls

No significant difference was found between the proportion of male partners and their selected controls with a positive GHQ score at any of the four assessment points (Figure 6.14). There was a tendency for male partners to show proportionately more distress at the time of receiving the female carrier's test result but this was shown not to be statistically significant (Chi-squared test p=<0.02). However, 14 of 15 (93%) partners with a positive GHQ1 had a female carrier partner who also had a positive GHQ score. Males were, therefore, significantly more likely to manifest psychological distress if their female counterpart was likewise distressed. (Chi-squared test p=<0.001).

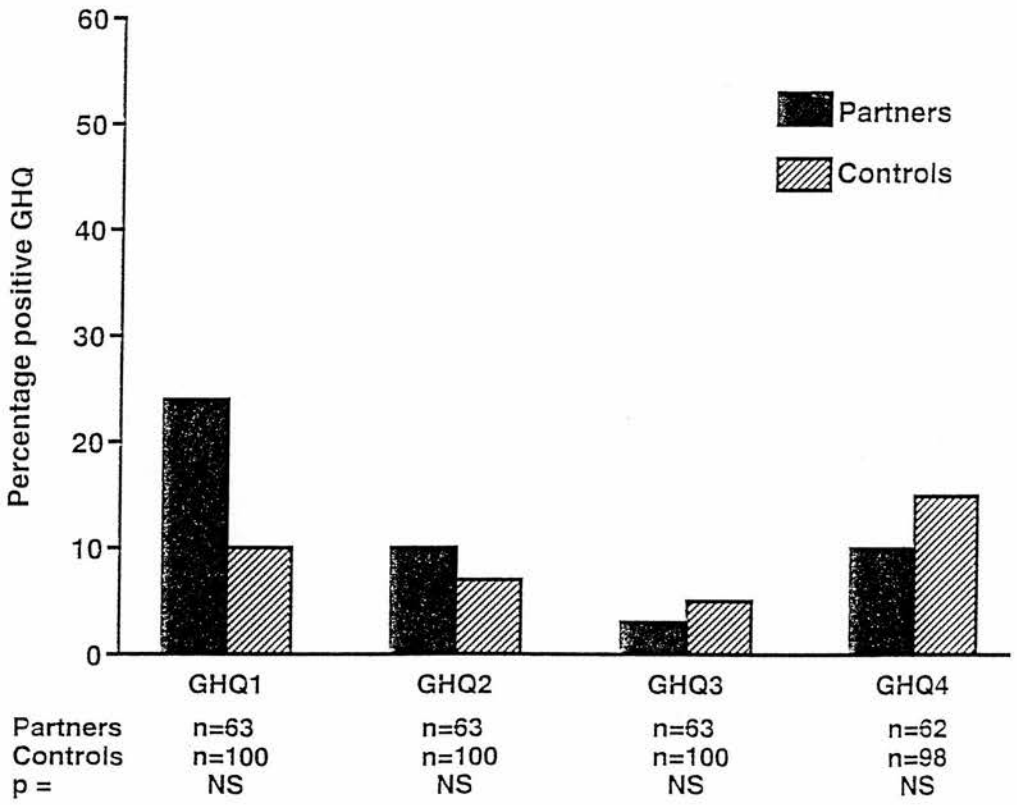


Figure 6.14 Male partners with a positive GHQ score compared to control subjects at each of the four assessment points

6.3.2.3 Results on the Symptom Rating Test (SRT)

Of the 576 women who presented at the antenatal booking clinic with a positive threshold GHQ 519 (90%) completed a threshold Symptom Rating Test (SRT). One hundred women who presented with a negative threshold GHQ were selected consecutively and asked to complete a threshold SRT for comparison (table 6.12).

Table 6.12 Median of threshold Symptom Rating Test (SRT) scores of bookers who presented with a positive GHQ compared to a cohort of 100 women with negative GHQ scores

Threshold SRT score	Bookers with positive GHQ	Bookers with negative GHQ	
total	13	4	$p < 0{\cdot}001$
anxiety	4	1	$p < 0{\cdot}001$
depression	4	1	$p < 0{\cdot}001$
somatic	2	0	$p < 0{\cdot}05$
inadequacy	4	1	$p < 0{\cdot}001$

Women with a positive threshold GHQ had threshold SRT scores well below those reported for psychiatric patients (Fava et al 1983; Kellner and Sheffield 1973). Those with a negative threshold GHQ score submitted SRT scores below those of normal subjects in previous studies (Cochrane 1980; Kellner and Sheffield 1973).

Fourteen (22%) carriers and 25 (25%) selected controls who presented with a positive threshold GHQ at the booking clinic completed a threshold SRT. There was no significant difference in the threshold SRT scores of bookers, carriers and controls. When carrier women received their positive test result (SRT1) there was a significant difference between carriers and controls in the total SRT score for generalised psychological disturbance (median test, $p = <0{\cdot}005$) and specifically in the sub scores for anxiety and depression (median test, $p = < 0{\cdot}001$). On receiving their partner's negative test result

(SRT2) the scores of carriers returned to control levels and remained there at the six week post-test point (SRT3) and again at the six week postpartum assessment point (SRT4) (Table 6.13).

Table 6.13 Median of Symptom Rating Test (SRT) scores of carriers and controls at each of the 4 assessment points

SRT1	**Carriers n = 64**	**Controls n = 101**	
total	11·5	7·0	p = <0·005
anxiety	4·5	1·0	p = <0·001
depression	4·0	2·0	p = <0.001
somatic	1·0	1·0	NS
inadequacy	2·5	2·0	NS
SRT2	**Carriers n = 64**	**Controls n = 101**	
total	7·0	7·0	NS
anxiety	2·0	1·0	NS
depression	2·0	2·0	NS
somatic	1·0	1·0	NS
inadequacy	2·0	2·0	NS
SRT3	**Carriers n = 64**	**Controls n = 101**	
total	5·0	7·0	NS
anxiety	1·0	1·0	NS
depression	2·0	2·0	NS
somatic	1·0	2·0	NS
inadequacy	2·0	2·0	NS
SRT4	**Carriers n = 64**	**Controls n = 99**	
total	6·0	7·0	NS
anxiety	1·0	1·0	NS
depression	2·0	2·0	NS
somatic	0·0	0·0	NS
inadequacy	2·0	2·0	NS

Partners did not complete a threshold SRT because only a proportion of males attend the antenatal booking clinics. There was no significant difference between later SRT scores for generalised psychological disturbance of the partners of carriers when compared with their selected controls. Anxiety and inadequacy sub scores were shown to be significantly higher than control subjects at the time when carriers were given their positive test results (median test, p = <0.05 and p = <0.02 respectively, table 6.14

Table 6.14 Median of Symptom Rating Test (SRT) scores of partners and controls at each of the four assessment points.

SRT1	**Partners n = 63**	**Controls n = 100**	
total	5·0	3·0	NS
anxiety	3·0	1·0	p = < 0·05
depression	1·0	1·0	NS
somatic	0·0	0·0	NS
inadequacy	1·0	0·0	p = <0·02
SRT2	**Partners n = 63**	**Controls n = 100**	
total	3·0	2·0	NS
anxiety	1·0	0·0	NS
depression	1·0	1·0	NS
somatic	0·0	0·0	NS
inadequacy	1·0	0·0	NS
SRT3	**Partners n = 63**	**Controls n = 100**	
total	2·0	2·5	NS
anxiety	0·0	0·5	NS
depression	0·0	1·0	NS
somatic	0·0	0·0	NS
inadequacy	1·0	0·0	NS
SRT4	**Partners n = 62**	**Controls n = 98**	
total	3·0	3·0	NS
anxiety	0·0	0·0	NS
depression	1·0	1·0	NS
somatic	0·0	0·0	NS
inadequacy	1·0	1·0	NS

There was a significant decrease in the sub scores of anxiety between SRT1 and SRT2 scores both in carriers and in their partners (Table 6.15). This was thought to be in response to the removal of threat to the fetus. Carriers similarly showed a decrease in the sub score for depression between SRT1 and SRT2 scores which was recognised as a subsidence of their feelings of loss of a normally progressing pregnancy.

Table 6.15 Comparison of median of SRT1 and SRT2 scores in carriers and partners

carriers n = 64	SRT1	SRT2	
total	11·5	7·0	NS
anxiety	4·5	2·0	p = <0·001
depression	4·0	2·0	p = <0·02
somatic	1·0	1·0	NS
inadequacy	2·5	2·0	NS
partners n = 63	**SRT1**	**SRT2**	
total	5·0	3·0	NS
anxiety	2·0	1·0	p = <0·005
depression	1·0	1·0	NS
somatic	0·0	0·0	NS
inadequacy	1·0	1·0	NS

6.3.3 Discussion

This study addressed the psychological effect upon 64 women who were told during pregnancy that they were CF carriers. These women had partners who received a negative screening test result and were reassured that it was unlikely that they were carriers. These couples were given a residual risk of 1 in 640 of having a child with CF.

There is an ongoing debate in relation to CF screening which centres on the population to be advised about the availability of the carrier test and whether screening should be concentrated on adults before pregnancy or pregnant women (Lipkin et al 1986; Brock 1984). Proponents of prenatal screening argue that pregnancy is the time when most individuals will be motivated to seek and use information on their carrier status But as new screening tests are developed and applied in pregnancy the psychological impact of their use must be considered. Understanding the potential emotional repercussions for women can help midwives offer enhanced support for mothers who are offered these tests.

Stress occurs when an individual perceives a threat that they will not be able to cope with. However, the influence of external factors must also be appreciated in the analysis of a stressful situation (Cochrane 1983). The threshold GHQ was used to detect stress at the time of booking before the screening test was carried out. Thirty two percent of women who were screened presented with a positive threshold GHQ. This was comparable with other studies in pregnancy (Sharp 1988).

It has been shown that randomly selected samples from the community will contain quite high proportions of persons with degrees of psychological disturbance ranging from mild to severe (Goldberg and Huxley 1992). Indeed it is estimated that 25 per cent of patients seen in general practice have anxiety as a clinically significant component of their condition (Wilkinson 1992) The GHQ revealed the value of using a questionnaire in the clinical setting. It proved an acceptable instrument which encouraged women to ventilate their feelings and could be considered for use as a quick and efficient method for the midwife to identify women who may require further help and support. Indeed, some have pleaded for psychological assessment to be an integral part of a woman's assessment in the prenatal period (Campbell and Field 1989).

Symptoms of early pregnancy was the most common reason given by women who submitted a positive GHQ score at booking (Table 6.10). *"Normally I am a confident, happy individual"* explained a 25 year primigravida who was a self-employed florist. *"Suddenly I have lost confidence, I am tired and emotional and find it difficult to make decisions."* Her husband was amused by this transformation in his generally assured wife, but she was plainly distressed about it. During discussion with the researcher it emerged

that she was worried that this was a permanent change and she would find it difficult to operate her successful business.

A substantial number of women were suffering from concurrent stress such as a poor obstetric history, difficulty conceiving, or an unplanned pregnancy. Older women were significantly more likely to present with a positive GHQ (Figure 6.10). This may be accounted for by the fact that women over the age of 35 years are more frequently faced with the prospect of invasive diagnostic tests for chromosomal abnormalities. The literature search revealed that older mothers may experience additional physiological and psychological stress because their age and experience make them more aware of the risks associated with pregnancy (Michelson and Gee 1984). Moreover, their pregnancy may be viewed negatively by others (Berryman and Windridge 1991).

Although no correlation was found between parity and a positive threshold GHQ, a poor obstetric history was cited by 10 per cent of women as the reason for their distress. The grief associated with an unsuccessful pregnancy is not a rare experience for women. In the UK one in a hundred babies die in the latter part of pregnancy or shortly after birth (Oakley 1984). Women are advised that spontaneous abortion is extremely common, occurring in around 1 in 6 pregnancies and in most cases the cause is not determined (Health Education Board 1993) Thus, many women are left asking the question "why". One woman interviewed for this study was convinced she had had 3 spontaneous abortions at approximately 8 weeks gestation. Because products of conception had been identified on only one occasion she perceived the medical profession were not convinced.

"I have a history of early miscarriage and I am off work and a little below par"

wrote this 32 year old social worker. On questioning by the researcher she revealed that she had *"taken to bed"* in an attempt to *"hold on to this pregnancy"*. She gave evidence

of the grief, mourning and despair that is experienced in early miscarriage; of feeling that she was denied the right to grieve; and that there was an apparent *"reluctant to acknowledge"* that she had a poor obstetric history because of a lack of pathological evidence. It is this unwillingness among health care professionals to award early miscarriage as a significant emotional experience which couples find so insensitive (Rajan and Oakley 1993).

Other women had completed an uneventful pregnancy only to deliver a stillborn baby. For many parents this loss was their first experience of death. The care of those who experience pregnancy loss varies greatly and support has been shown to make a dramatic and lasting difference (Rajan and Oakley 1993). Women in Rajan and Oakley's study felt a need for more information from doctors and for continuity of care. Also revealed was the need to be allowed to grieve in an emotionally-supportive environment. The fact that they experience awkwardness and a lack of understanding among relatives and friends requires that health care professionals do not appear unsupportive. Moreover, this study revealed that an appreciable difference in women's self confidence, and feelings of control over their lives which helped them cope in a subsequent pregnancy could result from midwives providing non-directive support. Although women entering the CF trial who had experienced infant loss felt they had recovered sufficiently to embark on another pregnancy, many stated that attending the booking clinic reawakened feelings of grief.

Some women were still adjusting to a loss or experiencing feelings of insecurity about the future of their subsequent pregnancy which one woman described:

> *"Well they said the chance of something like this (stillbirth) happening again would be very slight if any at all - but I think about it - I worry about it happening again."*

Women who had experienced an unsuccessful pregnancy reported feeling a need to have another baby: *"to reaffirm"* as one woman stated, *"my ability to bear a healthy child."*

A number had experienced therapeutic termination of pregnancy for fetal abnormality. Julie, a 21 year old and her husband had greatly felt the loss of their first baby at 19 weeks gestation when spina bifida was detected by maternal serum alpha-fetoprotein screening. A 35 year old couple who had 3 healthy children, had also experienced a therapeutic termination in their previous pregnancy when a fetal chromosome abnormality was detected. Both couples viewed the prospect of amniocentesis as a necessary and indeed welcome procedure to help ensure a healthy child but, as Julie and her husband stated:

> *"It changes your experience of pregnancy - you don't want to tell people just in case."*

The reluctance to disclose a pregnancy that may end in abortion has been revealed in other studies. (Rothman 1988). Women reported a reluctance to wear maternity clothes and kept their pregnancy 'private'. More disturbingly she found many women who underwent fetal diagnostic procedures were unwilling to form an attachment to their pregnancy. The trauma for these women when ultrasound scan visibly confirms the existence of their baby makes their denial more difficult and so they battle with their own instincts.

On a positive note, ultrasound scan offers reassurance to women feeling anxious about the viability of a pregnancy. One woman who presented with a positive threshold GHQ had suffered a perforated uterus following dilatation and curettage for a blighted

ovum. Having seen the fetus on the screen had reduced her anxiety and she expressed a rush of relief to the researcher afterwards:

"I feel so much better!"

Thirteen per cent of women cited an unplanned pregnancy as the primary cause for their psychological disturbance. Sarah, a 21 year old student explained:

"I'm feeling physically fine but I've been getting a lot of flak from my parents. I'd been travelling around the world and then came home to go to university, so my parents thought I'd finally settled down - then I got pregnant! My mother isn't speaking to me and can't understand why I don't want to live with the father of the baby."

A strained relationship with either the father of the baby or parents caused two per cent of women to present at the booking clinic with a positive threshold GHQ. A further five per cent stated this same circumstance as a secondary reason for their distress. For example, twenty eight year old Doreen had 3 live children and had experienced 6 spontaneous abortions. She wished to terminate her 13 week unplanned pregnancy. She stated she was *"struggling with a 1 year old son who has a food allergy, is sick a lot and doesn't sleep at nights."* She felt tired, irritable and ambivalent about the pregnancy. Her husband was opposed to abortion for the reasons Doreen had outlined but would countenance abortion for fetal abnormality. This couple were in agreement about undergoing all prenatal screening tests and perhaps Doreen perceived prenatal screening as a possible route to abortion.

Four per cent of women stated that having an active child at home was causing them to feel stress. It was a more common secondary reason cited by 9 per cent of women who submitted a positive threshold GHQ (data not shown). Having a child under the age of five years was found in one study to be the most important adverse influence on a

woman's mental health; more important than employment status or social class. Although young children had a profound effect on the mothers psychological health there was no significant effect on their physical health (Elliott and Huppert 1991).

This study commenced in 1991 and coincided with the Gulf War. Wives of soldiers from a nearby barracks booked to have their babies at the hospital. A number were anxious about their partners. Freda, a 23 year old felt bitter about her involuntary separation from her husband:

"Rick was sent out at the end of January and I got word the day I filled in the questionnaire that he won't be back until the end of this month. I feel really depressed about it - it's the third time we've been separated."

Two per cent of women were suffering from an existing illness. Karen, had myocarditis 5 years ago (4 months after the birth of her first baby). Since then she had suffered from myalgic encephalomyelitis (M.E.) and had recently experienced a relapse:

"I feel absolutely shattered - I have to get other people to do things for me. I want the CF test because I couldn't cope if I had a baby with CF."

Women experiencing stress frequently stated they found it easier to express their feelings by completing a GHQ and feeling encouraged to divulge their concerns during the interview. The SRT was particularly well received. Women said it helped them to formulate their feelings which some perceived as frighteningly unique. One woman said:

"I thought I was going mad - it's such a relief to realise that others must feel the same."

Another expressed her relief in the following terms:

"Having my feelings put into words - you know they exist - it's okay to feel them."

The psychological measures gave women a licence to admit feeling symptoms of stress and an opportunity to analyse and express how they felt. The pregnant woman may feel controlled by her pregnancy and not in full control of her life. A common occurrence was for women to present with a positive GHQ but subsequently submit a low SRT score. One reason for this may be that the GHQ acknowledges that women may feel stress during pregnancy and this they find reassuring. A second reason may be that talking to either the midwife or researcher helped decrease the focal stimulus.

Psychological assessment on all women screened served not only to ensure for the purposes of this study that there was no significant difference between carrier and control subjects at the outset, but proved valuable to the genetic nurse in the wider screening trial.

This study has shown that identifying a woman as a CF carrier during pregnancy does provoke a stressful response both in her and her partner. In 53 per cent of cases the identified carrier showed a significant increase in generalised psychological disturbance, specifically anxiety and depression, compared to 27 per cent of controls. This reaction occurred in response to learning of their carrier status and lasted for the period (approximately four days in this study) awaiting their partner's test result. It is likely that these women were responding to feelings of threat to the fetus resulting in anxiety, and to feelings of loss for a normally progressing pregnancy resulting in symptoms of depression. One carrier described her feelings thus:

> *"My husband kept telling me that statistically the odds were in our favour but I was convincing myself that I would probably end up having to have a termination. I think I was simply preparing myself for the worst."*

On receiving their partner's negative test result the distress subsided to control levels and remained there at all the other assessment points. Although the longer term effects are unknown, 12 (19%) carriers from this study have subsequently embarked on a further pregnancy.

Previous studies on patients undergoing prenatal screening have indicated that once a woman perceives her pregnancy has been threatened she continues to be concerned (Tabor and Jonsson 1987). The results of this study have shown a dramatic return to normal once a negative result is given. This corresponds with the other studies (Fava et al 1983; Burton et al 1985b; Tsoi et al 1987b). A probable reason for this is that stressful events may have a negative or positive consequence (Lennon 1989). Being identified as a CF carrier in the context of pregnancy implies a threat to the fetus. When the male partner receives a negative test result this is likely to be perceived as a positive consequence regardless of the residual risk to the fetus. Thus restitution ensues in the majority of cases. However, stress occurring during ongoing difficulties may cause delay in restitution (Goldberg and Huxley 1992) as experienced by 5 carriers who cited the presence of other life events as the major cause of their continuing psychological disturbance (Table 6.11).

Women who perceived their carrier risk correctly were less likely, though not significantly so, to present with a negative GHQ1 at the time of receiving their carrier result. This may reflect a more realistic perception of their partner's carrier risk. Previous studies suggest that women frequently accept prenatal screening tests to be reassured and are subsequently shocked when they receive a positive test result

especially when they did not consider themselves to be at high risk (Farrant 1985; Tsoi et al 1987b).).

Prospective anxiety about the screening test had no significant effect on carrier's GHQ1 scores at the time of receiving their positive test result. Women who had felt reassured about screening were as likely to manifest signs of psychological disturbance on learning they were a carrier as those who had felt anxious about being screened. The long term psychological impact on those receiving a positive carrier test result is as yet unknown. Loss of self-image and feelings of stigmatisation have both been cited as possible aftermath of heterozygote genetic screening (Kenen and Schmidt 1978). Will carriers view themselves as flawed? A number of empirical studies, mostly associated with Tay Sachs carrier screening, indicate that few individuals feel stigmatised or suffer a loss of self image on learning of their carrier status (Childs et al 1976; Clow and Scriver 1977). Despite the entity of 'gene carrier' being a normal fact of life it is likely to take time for the public to recognise this (Burn 1993). The dangers of public misinformation leading to stigmatisation are recognised (Goffman 1963). Indubitably, the consequences of this are conveyed clearly in the following text originating from a survey of genetic counsellors and nurses' views of CF screening in in The United States:

> *"Carrier screening can be a loaded gun; just this week one of our patients learned he was a carrier of the delta F508 mutation and his fiancee broke off their engagement. Now not only has he been dealt the bad news of being a carrier, his personal life is in a shambles and we have spent a great deal of time addressing his feelings of guilt, anger and betrayal."*
> *(United States, Office of Technology Assessment, 1992b)*

In contrast to male partners, SRT inadequacy subscores among carriers and their controls failed to detect a difference at any of the assessment points. A central theme throughout the trial during the pre-screening information and counselling stage and in

counselling carriers and their partners was the harmlessness of the single gene carrier state. Both verbal and written information given to carriers emphasised that they were no different from anyone else; that every individual carried altered genes; and that their health would not be affected. Nonetheless, follow-up will be required to evaluate if there are longer term consequences resulting from a woman learning of her CF carrier status.

Unlike most prenatal screening tests the CF carrier test involves the male partner. At the first antenatal visit an ultrasound scan allows the father early visual confirmation of the pregnancy which can initiate emotional involvement in the pregnancy (Pratt 1990). Pregnancy is a time when men too experience stress (Condon 1987). Thus it was considered important that this study focus on measuring the psychological impact of prenatal CF carrier screening on partners as well as carrier women. Male partners manifested symptoms of anxiety and inadequacy during the period awaiting their test result, but this disappeared on receipt of a negative test result. Both partners and male control subjects were significantly more likely to manifest psychological disturbance if their female partner was distressed. Condon (1987) studied the psychological and physical symptoms experienced by men and women during pregnancy. He found that fears about fetal abnormality was a factor which caused both sexes to manifest psychological symptoms. Marital insecurity was a factor which caused males alone to manifest both psychological and physical symptoms. Two men submitted positive GHQ scores throughout the CF study and both subsequently separated from their partners. As a result, one partner did not complete a GHQ 4 measure.

Almost without exception couples asked why they could not have been screened together at the time of booking to avoid awaiting the male partner's test result. Most

thought that on balance being given the test result prior to the counselling session was the correct procedure. One male partner voiced his opinion thus:

> *"It gave us time to talk and come prepared for the counselling session I feel it would have been a less productive meeting if you had only revealed the result at the time of the counselling session, whereas knowing in advance allowed us to think about what we wanted to ask - read the leaflet again."*

Genetic technology is making great leaps and it may only be a matter of time before it becomes possible to screen the population for other genetic disorders. Individual and social pressures to participate in screening programmes are considerable and a majority of women accept screening. There is a need for midwives to be able to assess and cope with the psychological effects induced by these tests in the context of pregnancy. This study has shown that for couples where the woman is identified as a CF carrier and her partner receives a negative test result the problems are minor and short lived. But, if a woman were to receive several positive screening tests during a pregnancy it could all too easily become a major focus for a considerable period. The need for midwives to appreciate and respond to the psychological needs of mothers undergoing these tests, is as great as the woman's need for them to be met.

A crucial question was whether women and their partners would feel on hindsight that pregnancy was a legitimate time to offer CF carrier screening and whether they had regrets over their decision to be screened. The decision to be screened could be affected by the way the test was offered, the accuracy of the information conveyed by the midwife, and her ability to do so in a way that is easily understood. The knowledge and attitude of carriers and their partners compared to selected controls was assessed. The results of this study will be presented in the following section.

6.4 Question 4: Do carriers and their partners understand the essential facts concerning CF carrier screening and what is their attitude toward having been screened?

6.4.1 The sample and methods

Between May 1991 and December 1991 a total of 69 women were identified as CF carriers through a prenatal screening trial. In all cases the male partner was screened. Three couples were identified where both partners carried a CF mutation and these were excluded from this study. In one case the pregnancy did not continue and one couple failed to complete the questionnaires. A further exclusion was a male partner who expressed reluctance to enter the study, however, his carrier partner wished to participate. A total of 64 carriers and 63 partners completed questionnaires.

Two control subjects were selected for each carrier. The controls had attended the same booking clinic as the carrier, were of the same parity and had received a negative test result. The male partners of these female controls were also invited to participate in the study, and served as controls for the partners of carriers. In all 116 female controls and 115 male controls agreed to take part, of whom 13 did not respond to the questionnaire, while a further two couples had pregnancies which failed to continue. A total of 101 female controls and 100 male controls completed the questionnaires.

With their booking clinic appointment all antenatal patients attending the hospital received a leaflet outlining the aims of prenatal CF carrier testing and describing the screening procedure. The leaflet stated a population carrier frequency of 1 in 25 and explained that carrier couples had a 1 in 4 risk of an affected child. The mode of inheritance of CF was described in diagrammatic form. The leaflet emphasised that a

family history of CF was not a prerequisite to being a CF carrier. Care was taken to stress the fact that being a carrier was unimportant unless the partner was also a carrier.

Women identified as CF carriers were invited to attend the hospital with their partner for counselling. An additional leaflet was given to all carriers and their partners reiterating in more detail the pre-screening information. This leaflet gave the risk of a carrier's siblings also being carriers; recommended that relatives should be screened prior to pregnancy; and advised on how relatives could initiate screening. Male partners received their test result after an average of 4 days. Six weeks after screening carriers and their partners were sent a "facts and feelings" questionnaire by post with a stamped addressed envelope for return.

Socio-demographic data was obtained from women's antenatal records. Questionnaire data was entered into a computer data-base for storage and analysis. Significance of results was assessed by the Chi-squared test.

6.4.2 Presentation of data

6.4.2.1 Characteristics of study population

Socio-demographic data is shown in Table 6.16. There were no significant differences between carriers and controls or between male partners and their respective controls in any of the factors examined.

Table 6.16 Characteristics of study population: carriers, partners and controls

	Carriers n = 64	Controls n = 101	Partners n = 63	Controls n = 100
Age (years)				
mean	27·86	28·64	28·13	30·38
range	18-44	20-40	18-46	20-48
Parity				
primiparous	35 (55%)	52 (51%)		
multiparous	29 (45%)	49 (49%)		
total	64 (100%)	101 (100%)		
Gestation				
mean	11·94	12·25		
range	7-16	7-18		
Marital status				
married	48 (74%)	82 (81%)	48 (76%)	82 (82%)
single	14 (22%)	16 (16%)	15 (24%)	17 (17%)
divorced	0	3 (3%)	0	1 (1%)
separated	1 (2%)	0	0	0
widowed	1 (2%)	0	0	0
total	64 (100%)	101 (100%)	63 (100%)	100 (100%)
Social class				
1	5 (8%)	11 (11%)	6 (10%)	11 (11%)
2	18 (28%)	31 (31%)	16 (25%)	31 (31%)
3	23 (36%)	36 (35%)	22 (35%)	36 (36%)
4	10 (16%)	16 (16%)	9 (14%)	16 (16%)
5	4 (6%)	3 (3%)	3 (5%)	3 (3%)
student	0	1 (1%)	0	1 (1%)
unemployed	4 (6%)	3 (3%)	7 (11%)	2 (2%)
total	64 (100%)	101 (100%)	63 (100%)	100 (100%)

6.4.2.2 Facts Questionnaire

There were six questions in the " facts" questionnaire. Tables 6.17 and 6.18 show the questions along with the responses. The correct response is highlighted.

Table 6.17 Percentage responses to "facts questionnaire"

Question	Carrier n = 64	Control n = 101	Partner n = 63	Control n= 100
1 Any couple can have a child with CF				
true	39	41	43	42
false	**61**	**59**	**57**	**58**
2 A couple can have a child with CF if:				
a) only one partner carries a CF gene				
true	11	10	13	18
false	**89**	**90**	**87**	**82**
b) if both partners carry a CF gene:				
true	**95**	**92**	**98**	**84**
false	5	8	2 *	16 *
3. One in 25 people in Britain carry a single CF gene				
true	**100**	**77**	**92**	**74**
false	0 †	23 †	8 *	26 *
4. Even if you have no family history of CF you can carry a single CF gene				
true	**91**	**94**	**89**	**96**
false	9	6	11	4
5. If both partners carry a single CF gene their chance of having a child with CF is: a) 1 in 2	2	0	2	4
b) 1 in 4	**98**	**94**	**97 ***	**81 ***
c) 1 in 20	0	6	1	14
d) all their children will have CF	0	0	0	1

Chi squared test on case versus control: * p = <0·01, † p = <0·001

Question 1 generated an unexpected range of responses which suggested ambiguity which had not become apparent in the questionnaire pilot study.

Responses to question 2a indicated that overall there was a good understanding of how CF was inherited. Carriers and male partners who answered "true" to this question added comments which revealed that the false negative rate of the test had influenced their response. During counselling carriers and their partners were told that if the male partners test result was negative they still had a residual risk of 1 in 640 of having a CF child. This information was also reiterated in a letter reporting the partners test result and in the patient information leaflet.

Question 2b showed the controls of male partners to be significantly more likely to misunderstand the inheritance of CF (Table 6.17). There was no difference in the proportion of male partners and their controls who had read the pre-screening information leaflet (71% and 72% respectively) as analysed in the pre-screening questionnaire (Section 6.1). However, an unknown number of males would have attended the antenatal clinic and may have benefited from pre-screening information and counselling from a midwife. Male partners, in addition, had undergone genetic counselling and had been given a further detailed information leaflet.

Both carriers and their male partners were significantly more likely to know the population carrier risk than their respective controls (Table 6.17 question 3). In counselling of carriers and their partners the population carrier frequency was emphasised because it equated with the partner's risk of being a carrier.

It was encouraging to find from the response to question 4, that the vast majority of subjects were aware that a family history was not a pre-requisite to being a carrier.

Responses to question 5 showed that male controls were significantly more likely than male partners to be mistaken about the risk of a carrier couple having an affected child (Table 6.17 question 5). Fourteen percent thought the risk was substantially lower (1 in 20) than the actual risk (1 in 4). This may reflect the absence among a number of male controls of pre-screening counselling received by their female counterparts at the booking clinic.

A particularly encouraging response was obtained to question 6 (Table 6.18) which indicated that, almost without exception, all subjects understood correctly the significance of carrying a single CF gene.

Table 6.18 Percentage response to "facts questionnaire" question 6

Question	**Carrier n = 64**		**Control n = 101**		**Partner n = 63**		**Control n = 100**	
If you carry a single a CF gene this means:	**true**	**false**	**true**	**false**	**true**	**false**	**true**	**false**
a) your health will be affected	0	100	0	100	0	100	1	99
b) you will develop the disease CF	0	100	1	99	0	100	2	98
c) it is only important if your partner carries a CF gene	**100**	**0**	**96**	**4**	**100**	**0**	**93**	7

6.4.2.3 Feelings Questionnaire

Carriers were significantly more likely than their respective controls to feel that the information given at the antenatal clinic was insufficient (Table 6.19 question 1b).

Table 6.19 Percentage responding to "feelings questionnaire" question 1

I feel that the information I / my partner was given about the CF carrier test a) before the antenatal clinic b) at the antenatal clinic was: 'about right,' 'too much' or 'not enough.'

a) before the antenatal clinic	about right %	too much %	not enough %
carriers n = 64	63	2	35
female controls n = 101	65	0	35
partners n = 63	62	2	36
male controls n = 100	54	0	46
b) at the antenatal clinic			
carriers n = 64	75	2	23 †
female controls n = 100	88	0	12 †
partners n = 63	87	2	11
male controls n = 99	87	2	11

Chi squared test on cases versus controls, † p =<0·001

A substantial number of male partners and their controls felt they had not understood the nature and purpose of the test before their female partner was screened (Table 6.20 Question 2).

Table 6.20 Percentage responding to "feelings questionnaire" question 2

I feel that I understood what the CF test was all about before I / my partner was tested.

	Yes %	No/don't know %
carriers n = 64	70	30
female controls n = 101	83	17
partners n = 63	54	46
male controls n = 100	60	40

Carriers were significantly more likely than their controls to have ambivalent feelings about having been screened (Table 6.21 question 3).

Table 6.21 Percentage responding to "feelings questionnaire" question 3

I am glad that I / my partner had the CF carrier test

	Yes %	No/ don't know %
carriers n = 64	80	20 †
female controls n = 101	97	3 †
partner n = 63	90	10
male controls n = 100	98	2
Chi squared test on cases versus controls, † p = <0·001		

Three carriers regretted having been screened and two had partners who felt similar regret. Notably, these two male partners submitted positive GHQ 1 scores and high SRT scores (SRT total = 48 and 38 respectively) Their scores were above those of their particular female partners. One carrier who expressed regret had a partner who stated he was " glad " she had been screened. Interestingly he, unlike the other two partners, submitted a low SRT score (SRT total = 7). Ten women answered *"don't know"* to question 3 of whom seven had male partners who stated they were *"glad"* they had been screened. Only one of their male partners expressed regret that his wife had been screened and a further two male partners felt ambivalent. Despite this, 88 per cent of carriers still felt that the screening test should routinely be offered to pregnant women (Table 6.22 quest 4).

Table 6.22 Percentage responding to "feelings questionnaire"question 4
I feel that the CF carrier test should routinely be offered to pregnant women

	Yes %	No / don't know %
carriers n = 64	88	12
female controls n = 101	96	4
partners n = 63	94	6
male controls n = 100	94	6

There was strong support for carrier screening in family planning clinics and GP health centres but less enthusiasm for screening in schools (Table 6.23 question 5).

Table 6.23 Percentage responding to "feelings questionnaire" question 5

I feel that I am in favour of testing for CF carriers in: a) schools, b) family planning clinics c) GP health centres

	Schools %	Family planning clinics %	GP health centres %
carriers n = 64	30	86	89
female controls n = 101	36	89	93
partners n = 63	32	81	87
male controls n = 100	38	81	89

Asked who they would tell if they found they carried a single CF gene, revealed that carriers and their partners were significantly more likely than their respective controls to tell their siblings and their children (Question 6.24 question 6).

Table 6.24 Percentage responding to "feelings questionnaire"question 6

If I turned out to carry a single CF gene, I feel I would tell my:

	partner %	my brothers and sisters %	children %	other relatives %	friends %
carriers n =64	100	92 †	92 †	59	45
female controls n =101	100	78 †	78 †	53	31
partners n = 63	100	79 †	87	49	30
male controls n = 100	100	63 †	79	34	22

Chi squared test on cases versus controls (agreement / non agreement); † p = <0·05

6.4.3 Discussion

This study attempted to assess two important aspects of prenatal screening for CF carriers; the understanding by the participants of the essential facts concerning CF carrier screening and their feelings about the delivery of screening information and having agreed to take part. The four groups involved had rather different access to information and also different experiences of the programme. Carriers and their partners would be expected to be more motivated in their attempts to acquire, comprehend and retain information.

Both carriers and their partners had been counselled in one-to-one sessions with a genetic nurse and had received additional written information. In contrast, the two control groups would have derived most of their knowledge from the information leaflet sent out at the time of their booking appointment, two to three months before they completed the "facts and feelings" questionnaire. Female controls received pre-screening counselling delivered by a midwife at the booking clinic but their male partner would only have done so if he had attended the clinic with her. It is therefore gratifying that in some sections of the "facts" questionnaire, all four groups had near maximum scores. This was seen in the response to whether it was possible to carry a CF gene if there was no family history of the disease (question 4, Table 6.17) and in response to the question about the effect of being a CF carrier on general health (question 6, Table 6.18).

Concern has been raised that being a carrier of a recessive gene may cause an individual to have a less positive view of his or her health (Marteau et al 1992b). In a study involving 27 carriers of the recessive gene that causes Tay-Sachs disease, 55 non carriers and 52 people who had not been screened, carriers were found to view their future health less optimistically than the other two groups. Prior to screening most

individuals do not consciously consider that they might carry any abnormal genes and take for granted that all of their genes are functional. "A new dimension to personal self has been actualised which may be perceived as defective, and for which no previous allowance in self and family concept existed" (Antley 1976 page 112).

Three of the four groups had an excellent idea of the risk of having a child with CF when both partners carried a mutant gene (question 5, Table 6.17). However, even though controls of the female carriers were well informed on this point, the male controls of their partners proved fallible. One reason may be that not all partners read the pre-screening leaflet (71% partners of carriers and 72% of male controls). A second reason is that whereas the partners of carriers received genetic counselling and a more detailed information leaflet, their male controls did not. Thirdly, male controls in contrast to the male partners had no reason to perceive themselves at risk and feel the need to understand the consequences of screening.

The fact that controls scored poorly in comparison to cases in the question about the CF carrier frequency in Britain (question 3 table 6.17) is not surprising. During counselling and in the additional information leaflet given to carriers and their partners this figure was stressed for two reasons. Firstly, to put the partners carrier risk into perspective and secondly, to emphasise the "normality" of the carrier status: *"Lots of us are CF carriers - 1 in 25 people."* Nonetheless, the first sentence of the pre-screening information leaflet stated that: *"One in every 25 men and women carry a single cystic fibrosis gene."* A similar finding was reported by Watson and colleagues in a CF carrier screening trial delivered through primary health care services (Watson et al 1992)

Responses to the "facts questionnaire" served also to evaluate the information leaflet. Approximately 40% of all groups thought the statement "any couple can have a child

with CF to be true (question 1, table 6.17 Although the leaflet showed in diagrammatic form that each partner had to be a carrier in order to have a child with CF, it also emphasised the incompleteness of the screening test: *" testing for the CF gene is still only successful in 85% of cases "*. Thus in a literal sense the statement is true, even though we expected respondents to find it false. This illustrates how easy it is to introduce ambiguities into a questionnaire and to fail to inspect in detail the content of printed information issued to patients.

From the "feelings" questionnaire, it would seem that there is a desire amongst all four groups for more information on the carrier test before attending the antenatal clinic (question 1a, table 6.19). During the designing of the information leaflet a study was carried out to assess the needs of the target population. Fifteen per cent of women thought the pilot leaflet should give more information and a further 11 per cent stated they did not know if additional information would be helpful. Of those who wished more information a majority indicated that more details about the disease CF was needed. A conclusion of that study was that regular evaluation of patient information leaflets would continue to be a basic requirement of the screening programme and that periodically the target population should be questioned about the acceptability of the information leaflets issued to them.

There was a significant difference between carriers and their controls in feelings about whether enough information had been supplied at the antenatal clinic (question 1b, table 6.19). Clearly considerable care is required to ensure women and their partners understand the nature and reasons for undergoing a particular prenatal test. From previous studies it is apparent that women commonly show quite major gaps in their knowledge of prenatal screening tests. (Donnai et al 1981; Faden et al 1985; Marteau et

al 1988a). In Donnai's study 12 women who had undergone therapeutic termination of pregnancy for fetal abnormality were found in most cases to have understood the reasons for the termination, however, a minority remained in doubt about the precise indication.

A much larger proportion of men than women did not feel that they knew what the CF test was all about before either they or their partners were tested (question 2, table 6.20). Again this may reflect the fact that approximately 30 per cent of men did not read the pre-screening information leaflet (section 6.1 figure 6.5 page 135) and an unestimated number did not attend the booking clinic. Moreover, the leaflet invited women to be screened not their partners, therefore males may have perceived themselves to be at less risk.

A large majority of all four groups were apparently glad that they or their partner had been screened (question 3, table 6.21). The only ambivalence appeared among carriers themselves. The stress experienced by receiving a positive CF test result influenced the attitude of 13 (20%) carriers among whom 3 women indicated they were *" not glad "* to have been screened and the remaining 10 answered *" don't know."* (question 3 table 6.21). Each of these carrier women scored positive on the GHQ1 at the time of receiving their positive test results, and 8 continued to score positive on the GHQ2, at the time of receiving their partner's negative test result. Positive GHQ results were submitted throughout the study by two of these women for reasons of recent bereavement one case, and attacks of breathlessness, one case. A further two women decided against maternal serum alpha-fetoprotein screening because of the stress experienced in undergoing CF carrier testing. Exactly how carriers perceive their newly discovered genetic status was not explored. Interviews would have perhaps gleaned a better understanding of women's attitude to this aspect of screening because nuances

such as quality of feelings and emotional overtones often emerge which cannot be conveyed by a questionnaire (Reynolds 1974).

It is interesting that despite 20 per cent of carriers indicating either regret or ambivalence about having been screened, only 12 per cent implied a similar view about routinely offering the test to pregnant women. There seems a considerable tolerance for the timing of the CF carrier test, with high proportions of all four groups feeling that it should be routinely offered in pregnancy. Screening through family planning clinics and GP health centres were supported by the majority of groups. There was less enthusiasm for screening in schools in contrast to other studies in which this was cited as a popular option (Green 1992; Zeesman et al 1984). However, the consequences of screening in any one of these situations and the decisions and actions which might emanate were not outlined to participants, therefore, they may not have considered these when responding.

Some are of the opinion that divergent approaches to carrier screening are likely to be complementary (Burn 1993). The evidence from previous genetic screening programmes is that the antenatal booking clinic are likely to be the ultimate option because patients and physicians frequently regard genetic screening as part of reproductive care rather than reproductive decision making (Shapiro and Shapiro1989).

All respondents stated that they would tell their partner if they turned out to be a CF carrier (question 6, table 6.24). Carriers and their partners were more likely to tell their siblings, children, other relatives and friends about being a carrier than were their respective controls. This finding is likely to be as a result of the one-to-one counselling sessions with the genetic nurse during which a family pedigree was drawn out and risks to siblings explained. In addition the information leaflet issued to carriers outlined the screening procedure for relatives.

Data concerning the number of male controls who attended the booking clinic was not collected making it impossible to assess whether this influenced knowledge and comprehension. Nonetheless, the response from this group was extremely encouraging with a majority answering the questions correctly. Criticism has been levelled by male partners at the apparent lack of interest taken in them by obstetric staff (Jordan 1990). The high level of comprehension by this group perhaps reflects the fact that the CF test could potentially involve them and accordingly they were interested. It may also support the concept that people accept preventive measures when they perceive the disorder as serious, their being susceptible and that there are benefits (Childs et al 1976).

This study has shown that firstly a simple carefully designed leaflet can be one effective way of communicating essential facts about prenatal screening, especially when it is supported by counselling by a midwife. Both the leaflet and counselling had been issued several months before the control subjects answered the questionnaire. Although all female controls were also given a great deal of information on a variety of topics during the booking-in procedure, in both verbal and written form, they apparently retained and understood most of the information on CF carrier screening.

Although all four groups in this study had a favourable attitude toward CF carrier screening in pregnancy such screening needs to be accompanied by educational endeavour and sensitive application. All four studies presented in this thesis show how genetic screening generates a complex range of ethical issues which confront individuals, families and professionals. These and other aspects essential to the judicious and sensitive application of prenatal CF screening by midwives are explored in the overall discussion presented in the following chapter.

CHAPTER 7
DISCUSSION

Discussion

A strength of the research carried out in this thesis lies in the large sample size which helped to clarify women's perceptions of screening and to quantitatively analyse their responses in relation to a new prenatal screening test. The integration of qualitative and quantitative research methodology enhanced the validity of the study findings by adding a perspective that numbers alone could not have provide. Qualitative research clarified important concepts through the exploration of pregnant women's individual perceptions and personal reactions to the offer of prenatal carrier screening for CF. While facts and figures were provided, the main concern was to convey the feelings, concerns and experiences of women exposed to a recent development in medical technology and to look beyond and consider the implications for midwives. At a time when midwives are extending their skills to implement continuity of care which will facilitate the full involvement of mothers in exercising choice (Scottish Home and Health Department 1993), the findings of the research presented in this thesis are especially germane. "The motive for carrying out nursing research is not just to gain knowledge but to influence what is practiced as a result of what is learned" (Abbot and Sapsford 1992 p viii).

As additional prenatal screening tests become available and as society grapples with the increasingly complex questions raised by the new techniques which provide the means for these tests, individual women and couples are left to make their own particular decisions. The findings in this thesis suggest that around 30 per cent of the women rely upon the midwife to help them decide whether or not to be screened. Midwives are confronted by ethical issues relating to the care of mothers and their babies at all stages antenatal, intranatal and postnatal, but the new technology which facilitates an increasing number of prenatal screening tests poses yet more potential ethical dilemmas. To attempt

to identify and explore all the ethical issues relating to prenatal screening is not possible in this text. There are, however, a number of ethical considerations which thread through the whole screening procedure and warrant exploration.

7.1 Ethical aspects of prenatal screening for CF carriers

Ethical issues in relation to genetic screening have been reported by the Nuffield Council on Bioethics (1993). The report bases a considerable amount of its findings on published excerpts from the results of this thesis. The committee acknowledge that they could not hope to identify and attempt to answer all the ethical questions which could confront individuals, families and professionals. There are additional issues which the studies in this thesis expose and which will be addressed here. Some apply solely to prenatal genetic screening and others are common to genetic screening per se.

Ethics has been defined as "the study of the underlying reasons for deciding what is best in the face of conflicting choices" (Wilday 1989 page 176). The principal reason for addressing ethical issues raised by prenatal CF carrier screening relates to the future care of individuals who are invited to be screened. Most research involves small numbers of subjects which permit attention to important ethical details. Experiences from previous genetic screening programmes show that fundamental issues such as confidentiality, autonomy and consent can be neglected when research becomes service (Rowley 1984; Roberts 1990). For example, public education, adequate counselling and post-screening support are necessary infrastructures without which individuals can all too easily misinterpret the meaning of a positive screening result (Whitten 1993). Written consent to be screened is frequently abandoned once research becomes service. In the hospital in which the prenatal CF carrier screening trial took place, women were not asked for signed consent in order to undergo maternal serum alpha-

fetoprotein screening. A further concern was that the hospital leaflet outlining MSAFP screening did not a) draw attention to termination of pregnancy as a possible consequence of screening nor b) mention that the test could only detect 85 per cent of pregnancies at risk of neural tube defects and 60 per cent of those at risk of a chromosomal anomaly (Cunningham and Gilstrap 1991).

The aim of this discussion, about ethical issues raised by the research findings presented in this thesis, is to highlight a number of ethical problems posed for midwives and for the women they care for in relation to prenatal CF carrier screening. Questions will be raised rather than necessarily answered.

7.1.1 To screen or not to screen

"Cystic fibrosis illustrates a moral paradox within medicine which on the one hand seeks to screen and eliminate through abortion and on the other to treat and cure" (Elborn 1991 page 40). Current treatment for CF and improving life expectancy, coupled with the possibility that gene therapy may also improve the quality of life of sufferers, leads to the argument that financial resources should be directed toward therapy rather than carrier screening programmes (Elborn 1991). Conversely, screening could be justified on the grounds that gene therapy is likely to be expensive, and that lowering the incidence of CF could directly benefit surviving sufferers by reducing the total number who stand to benefit from therapy. In the final analysis the relative benefits and risks of gene therapy compared to the burden and prognosis of the disease may determine the future of screening.

A response to population genetic screening is that it could cause the public to view CF as a disease which should disappear and, therefore, they might consider targeting money toward better treatment inappropriate (Wilfond and Fost 1990). Do the results of

the studies in this thesis help answer the question: should society choose therapy in preference to screening?

Participants in the prenatal CF screening trial chose to undergo prenatal carrier testing for a variety of reasons (Chapter 6, page 149, table 6.6). Sixty two per cent of women wished to be screened to find out during pregnancy if the baby had or was at risk of having CF. Of these women 55 per cent were not pursuing the option of terminating an affected pregnancy, indeed 10 per cent clearly stated that their reason for being screened was to prepare for the birth of an affected child. These women gave testimony to prenatal screening being perceived as of significant benefit to mother and child, even when the detection of an abnormality would not lead to termination of pregnancy.

One woman explained:

> *"As I understand the severity of the disease, I would like to know whether my child or future children were at risk and even if I decided against termination I would be mentally prepared for a child with CF."*

The worry of fetal abnormality can place a significant psychological burden on those who have experienced it in a previous pregnancy:

> *"I really want to know more about the baby I am expecting. My previous baby died at 3 days with an abnormality of the heart."*

An argument in favour of detecting a fetal condition prenatally is to allow perinatal or neonatal medical intervention which would benefit the infant (Clark and De Vore 1989). Ten percent of infants with CF are born with meconium ileus, a life threatening condition requiring prompt neonatal therapy (Goodchild and Dodge 1985). Yet, there is another very obvious direct benefit; the psychological and practical

preparation of parents for the birth of a child with special needs. As yet no studies have been carried out to reveal if there are measurable short and long term benefits to parents, affected children and unaffected siblings in families where the results of prenatal screening and diagnosis facilitate anticipation of a child with special needs. But, evidence suggest that women and their partners connect prenatal screening for CF with preparation for the birth of a child with special needs. A study of 135 couples who accepted prenatal CF carrier screening revealed that 91 (67%) felt that gene therapy would not influence their decision to be screened. Equal numbers, 21 (16%) couples stated they would either decline to be screened or felt ambivalent toward screening. Even if the life expectancy of CF sufferers increased to normal 78 per cent stated that they would wish to avail themselves of prenatal CF carrier screening (Mennie et al 1994). The desire to have information abut the baby before birth is a compelling reason for being screened. As one couple stated:

> *"We think we would still opt to be tested for CF even if gene therapy was available. Our reasons for having the test would be different though. If the quality of life and extended life expectancy could be improved, the question of whether or not to continue the pregnancy would no longer be a consideration, rather that when the child was born we would be prepared for it to be ill and have to undergo treatment. This would be much less distressing for parents than finding out after the birth."*

Although the primary goal of a screening programme is often perceived as a means to promote informed choice and to prevent suffering whether it be physical or psychological. It should never be organised in such a way that the birth of a CF child is seen as a "missed prevention" (Van den Berghe 1987) or as the result of irresponsible reproductive decisions (Wilfond and Fost 1990). The midwife can ensure that women understand that they will not experience coercion to end a pregnancy at risk of a child with special needs. There is a danger that screening may foster the attitude that a less than perfect fetus should be prevented (Curry 1994).

A further concern revolves around the potentiality of genetic screening becoming a women's issue (Chapple 1992). Because new developments in health care depend upon financial resources it may prove impossible to fund screening to the extent that it could be offered to all sections of the childbearing community. If some form of rationing were to be necessary then an obvious screening strategy would be a prenatal approach. This could be argued on the basis that outwith pregnancy money could be spent on screening individuals who may never use the information. If, however, screening were to focus on pregnant women it could put both the burden of responsibility for genetic disease and the emotional costs of screening on their shoulders. Thus choice should not simply involve whether to accept or decline the offer of CF carrier testing but when the offer is made.

7.1.2 Physician versus patient perception of screening

Clearly some women perceive prenatal screening as a means whereby they can prepare for the birth of an affected child. Immediately, this raises issues concerning physician versus patient perception of prenatal testing. Among obstetricians there are those who hold the view that it is inappropriate to carry out a risk associated procedure such as chorionic villus sampling or amniocentesis in a continuing pregnancy. They may advocate discouraging a couple opposed to termination from pursuing this option (Crawfurd 1983; Thorp and Bowes 1989). Now this could arguably place a midwife in a delicate situation. Given that prenatal CF carrier testing is presented to women by the midwife, she is placed between the patient's wishes and the obstetrician's opinion and this situation is not without its difficulties. The midwife is not free to take the same initiative as the obstetrician, nonetheless, she may feel she has a better understanding of what is, or is not, in the patient's and unborn child's best interest. While the obstetrician will look to the patient's and unborn infant's medical welfare, the midwife may look more to their general well-being (Sutton 1990). The Royal College of Physicians state that prenatal diagnosis should be available to women who

are completely opposed to termination of pregnancy, *"since testing may provide welcome reassurance, or an informed choice to care for a child with a known handicap"* (Royal College Physician 1989 page 49:8.6).

In a related vein a 25 year old para 1 stated:

> *"If CF is detected at this early stage, I believe it is better than finding out at birth."*

Regarding midwives' professional responsibility The Royal College of Midwives (1981) state that "in making a professional judgement (the midwife must consider) certain factors: the professional code which demands that the midwife must not harm the mother or baby; the primary responsibility of the midwife to use her knowledge, skill and power to promote the well-being of the mother and baby."

Some perceive the advantage of screening as its capacity to allay worry and for those who do not gain reassurance, advance knowledge provides the opportunity to prepare themselves emotionally, physically and financially for the challenges that lie ahead. Although there is a pregnancy loss-related risk associated with prenatal diagnostic procedures this may seem acceptable to some couples at a 1 in 4 risk of having a CF infant, particularly if they perceive there are potential benefits in having prior knowledge about the status of their unborn child.

Ideally, midwives and nurses should provide information and care in unison with other health care professionals. Clearly, there may be situations when the midwife will be obliged to initiate discussion, to take action and to raise objections in order to safeguard the interests of the mother and infant (Sutton 1990). Increasingly, women themselves are demanding more attention to their emotional needs in pregnancy, which

corresponds with "an important change in emphasis from need-led to demand-led health care." (Holland and Stewart 1990 chapter 1, page 6).

7.1.3 Access and demand

Resource constraints

"Individuals must have the right to choose whether or not to be tested and their choice will depend on the provision of complete information about the test and particularly its consequences" (Haan 1990 page 177). Patient autonomy is a recurring theme (Hodgkin and Yoxen 1985; Kings's Fund Forum 1987; Colten 1990; Sutton 1990; Weatherall 1991a; McGregor 1990). An individual's choice not to be screened must be respected and no pressure exerted. Given the increase in demand and the increasing scarcity of resources some are concerned with the question - who is entitled to prenatal diagnosis? (Fletcher and Wertz 1992). In other words, rather than there being a danger of patients being coerced into prenatal screening and diagnosis there is a danger that they may not be offered this option. Should CF carrier screening become an established part of pre-conceptual and prenatal care? Can regional health authorities afford to fund it? Could couples who failed to be offered screening sue for malpractice if they had a child with CF?

The Royal College of Physicians recommend that genetic screening and prenatal diagnosis should be equally available to the whole community as an established part of maternity care (Royal College Physicians 1989). Yet, currently screening for fetal abnormalities varies considerably from one area of the United Kingdom to another (Holland and Stewart 1990). Concern has been expressed that co-ordination, organisation and evaluation is not ideal (Holland and Stewart 1990; Cuckle 1990). A national screening body has been proposed to correct this situation (Cuckle 1990).

Professional ignorance.

Problems of access to screening may arise from poor organisation, limited professional and public awareness, and severely limited technical, educational and counselling resources. Shortcomings can be blamed to some extent on underfunding but missed opportunities arise because basic clinical genetic knowledge is lacked by both the medical and nursing profession (Royal College Physicians 1989). If an available prenatal test is not offered could this be cited as a cause of damage to the fetus and constitute grounds for litigation? The right of the child to claim damages was accepted in the United States when parents who underwent carrier testing for Tay-Sachs disease claimed the tests were negligently carried out resulting in their infant being born with the disease (Sutherland 1990). However, a UK case in which a child claimed damages on the grounds of failure of the mother's physician to advise her of the possible consequences of exposure to rubella was rejected, on the grounds that life resulting insevere disability was better than no life (Sutherland 1990).

Again, in the United States there was refusal to permit recovery of damages where a woman in her late thirties failed to be offered amniocentesis resulting in the birth of a child with Down's syndrome (Sutherland 1990).

What if a mother declines a prenatal test? Mothers have been held liable where accidental injury to the fetus resulted from her driving a motor vehicle. In such a case the insurance company settles the claim. However, it is difficult to envisage a child claiming damages as a result of the mother declining the offer of a prenatal screening test. Quite simply who would pay compensation? A more conceivable situation is that of alleged professional negligence, resulting in failure to provide or offer a test.

7.1.4 Issues of informed consent

Providing information

Genetic carrier screening can generate information about one individual which may well have implications for other related family members. Therefore, consent to be screened differs from consent to undergo an operation or other treatment. Moreover, screening is a process usually initiated by health care professionals. Ensuring that the information is given is not sufficient, the midwife should present it in such a way that women and their partners can appreciate the choices, and can make a decision which they will not regret in future. The midwives' role, in this context, can be viewed as that of a teacher.

Currently individuals in the prenatal CF screening programme are provided with information by means of a leaflet and a one-to-one counselling session with a midwife. As the pre-screening study described in chapter 6.1 revealed, 40 percent of women in the 16 to 20 year age group had not previously heard of CF (figure 6.2) and almost 16 percent of them found the leaflet difficult to understand (figure 6.3). This was reflected in the fact that 70 percent of this age group either perceived their carrier risk incorrectly or had no perception of their risk (figure 6.6). It is important that written and verbal information is in a language appropriate to the individual (Nuffield Council on Bioethics 1993). In practice the mode of delivery may be just as important. Many young people today are perhaps more familiar and consequently more comfortable acquiring information from videos. Studies indicate that a videotape containing the same information given in conventional counselling can be an effective method of providing pre-screening information when it is followed by an opportunity to question a health care professional (Fisher et al 1981; Rowley et al 1984). Research comparing the retention of information and appeal of

this method of imparting information to younger women and their partners could be valuable.

A recent review of Down's syndrome screening in pregnancy revealed that information provided is either not always adequate, or not retained. Consequently, women are not always sure what tests they have undergone and what the results mean (Statham and Green 1993). Continuing evaluation of written information, verbal information giving and counselling should be an integral part of any genetic screening programme.

Individuals with special needs

A not uncommon problem is providing information to women who have a learning difficulty, are deaf or blind. This problem is not unique to prenatal screening, it can pose an obstacle to providing general pregnancy care. Nine women in the trial were excluded because they had severe learning difficulties. The decision was made in all cases by the obstetrician. But who should ultimately make this decision? The problem of genetic testing of the mentally ill and those with severe learning difficulties has been addressed (Nuffield Council On Bioethics 1993). They state that: "it is a matter of consideration whether genetic tests on mentally ill individuals or those with severe learning disabilities should be permitted in situations where the information gained would be of clear benefit to other family members." Who does prenatal carrier screening for CF benefit? The benefit is to the mother and the child. Does an obstetrician have a right to withhold screening? Usually a relative, or surrogate decision-maker such as a close friend may decide. Alternatively help may be obtained through the social work department, or a court appointed legal guardian (Meyers 1990).

How much information should be provided

Studies indicate that women frequently enrol in antenatal screening programmes without being fully informed about the facts and issues (Marteau and Slack 1992a). There is a tendency, particularly during pregnancy, to sanction information which is non-confrontational and censor that which could be construed as alarmist. For example, during the designing of the CF pre-screening leaflet the original wording with respect to a female carrier's partner receiving a negative test result read: "If your partner's test result is negative then your risk of having a baby with CF is low." Obstetric staff wished this to read: " your risk of having a baby with CF is very low." As a compromise the word 'very' was not underlined! The way statements are phrased by health care professionals about reproductive risks can influence an individual's perception of the degree of risk (Shiloh and Sagi 1989).

A failing of many information leaflets is that they classify prenatal screening tests along with prenatal diagnostic tests. "Tests to detect abnormalities in the baby" (Health Education Board for Scotland 1993). It is interesting that no attempt is made to differentiate between the two. This is indeed a pity because it might go some way to reducing the stressful reaction which most women experience when told they have a positive screening test result. Nowhere is it explained that a screening test is merely an indicator to carry out a further test and not a fetal diagnostic test. In other words a screening test does not necessarily reflect the health of the baby. Both in writing and during verbal pre-screening information and counselling the midwife should specify the difference between a prenatal screening test and a prenatal diagnostic test. Between 3 and 5 percent of women will receive a positive maternal alpha-fetoprotein test result, but only 1 in 20 of these women will finally receive a positive prenatal diagnostic result and be faced with the decision to continue or end the pregnancy.

There is a danger that opportunities may be missed to educate pregnant women simply because health care professionals are inclined toward believing that too much information is not beneficial to the individual. Frequently it is perceived to alarm or confuse, but pregnant women are healthy and usually highly motivated individuals keen to learn all they can about their unborn child. All too often fear of the unknown is the cause of anxiety and this could arguably be alleviated by additional and accurate information.

The decision to be screened must remain that of an individual woman and her partner. But, should the amount of information given be left entirely to the wishes of individuals concerned, or should the midwife endeavour to impose as much information as she deems necessary for a woman to make an informed decision? For example, some women attend the booking clinic having failed to read the pre-screening information leaflet and show reluctance to address the counselling offered by the midwife. Despite the midwife emphasising the importance of appreciating the pre-screening information, some prove resistant. Yet, should they receive a positive test result they may express regret at having been screened.

Studies show that it is all too easy for women to receive a positive screening test result and only then realise they did not understand the implications of the test. A recent study on the effects of Down's screening during pregnancy reported that one woman did not read the information leaflet explaining serum screening for Down's syndrome, assuming it to be about screening for spina bifida (Statham and Green 1993). This study does not state whether participants signed a consent form.

It is easy to level criticism for misconceptions of screening tests at both staff and patients. What is required is a system which will help ensure that such problems are not

encountered. Signed consent may offer an important contribution to ensuring an informed decision is made about having initial screening tests.

Signed consent

There is strong argument for ensuring that a consent form is signed before any prenatal screening test is carried out. Indeed there is good reason to propose that separate consent be obtained for individual screening tests. Provision for this could be made in a booklet which outlined each screening test, and included separate consent forms for each. A consent form would be signed only after the midwife had explained a particular test and ensured a woman had both understood and had elected to have a test.

An additional safeguard would be to record those tests accepted and those declined on a woman's antenatal liaison card. This would ensure that the general practitioner was acquainted with the tests his patient had undertaken or wished to undertake. It might also function as a valuable record for the woman herself providing she was made aware that the information was recorded. During the prenatal cystic fibrosis screening trial an infant was admitted to the local paediatric hospital with symptoms indicative of cystic fibrosis. The mother informed the hospital that during pregnancy she had undergone cystic fibrosis carrier testing. When the woman's antenatal records were checked it was clearly documented on the copy of her liaison card that she had declined CF carrier screening, but had accepted maternal serum alpha-fetoprotein screening. A further check was that there was no signed consent form for the CF carrier test in her antenatal records. It could be argued that had signing a consent form been a mandatory procedure for maternal serum alpha-fetoprotein screening she would have been more aware of which tests she had chosen and which she had declined.

7.1.5 Issues of confidentiality and autonomy

Genetic screening may result in information generated about one individual in a family having implications for other family members. Genetic screening tests rely on nucleated cells from which DNA is derived. A pertinent issue raised revolves around the ownership of genetic material (Pullen 1990). Does this belong to the donor or to the laboratory who carried out the test? In relation to samples obtained during the prenatal screening trial it is believed that these are owned by the local health board.

Results of DNA analysis are filed in the patient's antenatal records. Carrier results are reported to the individual concerned, their general practitioner and obstetrician. No attempt is made to follow up relatives at risk. The individual carrier is informed during counselling, and by way of a carrier information leaflet, that any siblings have a 50 per cent chance of also being CF carriers. Details of how siblings can initiate screening are provided in the leaflet.

Autonomy of the fetus and the woman in genetic screening can be diametrically opposed. Some women clearly perceive that bringing up a child with a genetic disorder such as CF could have a devastating effect upon their own and their families life and for this reason will opt to terminate an affected pregnancy. The autonomy of the fetus is not, however, best served by abortion. Some predict future alternatives to termination of pregnancy which is currently a central issue of prenatal genetic screening. Rather they foresee prenatal genetic screening being applied to diseases where preventive therapy is possible for example, diet and cholesterol lowering drugs for ischaemic heart disease, early diagnosis and treatment of diabetes, carcinoma of the colon and breast (Bell (1990). Bell also identifies embryo selection for monogeneic disorders as an additional important option in the screening process.

A minority (9%) of women chose to be screened in order to establish their own carrier status, whereas 62 per cent sought information about the fetus (Table 6.6). Further evidence of the lack of concern they show for themselves is manifest in the psychological assessment of carriers where restitution occurs after receiving their partner's negative test result (Figure 6.13). Because the focus of concern is not upon their own individual carrier status it is unlikely that the implications to other family members are seriously considered. Indeed it is arguably unfair for health care professionals, relatives, or society to expect couples who undergo prenatal genetic screening to calculate this aspect of genetic screening and to allow it to influence their screening decision. Consequently, cascade screening, where relatives at risk are systematically contacted and offered screening, should perhaps be restricted to families where screening has been offered pre-conceptually.

7.1.6 Prenatal diagnosis

The attitude of health care professionals toward pre-natal diagnostic techniques may also influence how the benefits of chorionic villus sampling versus amniocentesis are conveyed. There are a number of perceived benefits in the choice of chorionic villus biopsy as a prenatal diagnostic procedure for a disorder such as cystic fibrosis; the laboratory analysis is straightforward; the procedure is carried out in the first trimester facilitating termination of pregnancy at an early stage. Counsellors frequently offset the higher risk of spontaneous abortion against a 1 in 4 risk of CF occurring in the fetus. However, the advantage of undergoing amniocentesis at a later stage of pregnancy is the lower risk of spontaneous abortion (less than 1%) (Lilford 1991). Some women may favour second trimester prenatal diagnosis by amniocentesis despite the possibility of a late termination of pregnancy.They may feel that knowingly placing a possibly healthy fetus at risk of spontaneously aborting through first trimester prenatal diagnosis is unacceptable. For those who wish to postpone

their decision until the status of the fetus is known, second trimester prenatal diagnosis may be preferable. It is only honest to highlight the advantages and disadvantages of both procedures when counselling couples at risk of having an affected child and providing there are no obstetric contra-indications to performing one or other procedure. The final decision should be that of the parents.

There is currently no dispute that parents have a choice to continue or to end a pregnancy where the fetus is affected by CF. But, regardless of their decision, are there adequate resources to support them? Post termination support is often poor, and can result in long term psychological sequelae (White-van Mourik 1992). By the same token the provision for coping with an affected child will vary from one area of the country to another. The Royal College of Physicians state that couples should never be pressed; on the other hand they caution that any decision should take into account the long term implications, such as care of the affected individual after their deaths (Royal College Physicians 1989). If a couple have strong moral principles then this will almost certainly regulate their decision to continue or end a pregnancy. Other couples will base their decision on the perceived effect of an affected child on family and social life, financial costs, and the problems of caring for the child at home (Wertz et al 1984).

The process of information giving and receiving can be impaired by the reactions couples experience in the coping process (Pullen 1990). Counselling for prenatal diagnosis has other goals, for example, the reduction of guilt, shame and anxiety as well as helping couples adjust to the presence of a genetic disorder (Wertz 1984).

Prenatal diagnosis can be viewed as having a pro-life effect because couples who might previously have avoided a pregnancy because of a known genetic risk may be more willing to conceive.

7.1.7 Evaluation of screening

It is ethically undesirable to evaluate a screening programme by cost-benefit analysis which measures effectiveness by the number of affected pregnancies which are terminated and the savings made from reducing the incidence of the disease. Nonetheless evaluating screening in monetary terms has been carried out (Gill et al 1987; Wald and Cuckle 1988).

A more acceptable method is by patient satisfaction (Shiloh et al 1990). Reservations have been expressed by Clarke (1993) about the feasibility of quantifying the evaluation of qualitative aspects of screening; work in this thesis indicates that this is possible. The feelings questionnaire was designed to assess satisfaction with information and counselling and in addition participant's attitude to pre-natal carrier screening. A simple questionnaire which also offered the facility to express individual opinions may not be perfect, but it can expose areas of dissatisfaction which may subsequently be comprehensively examined.

The recent development of population genetic screening for carriers of cystic fibrosis is only one of a number of recent developments in recombinant DNA technology. Increasingly these developments are impinging on society and gradually the community will become more aware of the role of genes in disease and of the implications of being able to identify susceptible individuals. As these new technologies are implemented, midwives should give careful consideration to the ethical issues which arise so that maximum benefit can be obtained for the individuals for whom they care as well as the wider community.

7.2 Implications for midwifery

The conceptual model of stress, coping and outcome (Cochrane 1983) contributed to the framework used to develop the studies which constitute this thesis. The model also provides a framework to assess how the results of the studies contribute to midwifery and nursing practice.

The results show that women who were offered cystic fibrosis screening during pregnancy were confronted by problems that were psychological rather than physical. Psychological adaptation is important not only in terms of the well-being of the pregnant woman herself (Watson et al 1984), but the eventual well-being of her baby (Newton et al 1979; Berkowitz and Kasl 1983; Muylder 1989). Using the conceptual model as a framework the results of the studies can be summarised to contribute to a knowledge base for the effective, judicious and sensitive presentation of screening, as well as examining areas where knowledge is lacking and where further research is indicated to understand these dimensions of prenatal genetic screening (figure 7.1). The results of the studies allow the researcher to suggest guidelines for the presentation and care of women who are offered prenatal genetic screening. Although these are consistent with presenting genetic screening within an antenatal clinic setting, they could equally apply to screening presented within an alternative setting, for example, in the community by health visitors or practice nurses.

7.2.1 Preparation for screening: a basis for coping

The pregnant woman's interpretation of events

Knowledge, attitudes, beliefs, expectations and predictions all contribute to a woman's appraisal and attitude to the screening test. The immediate stressor in this study was

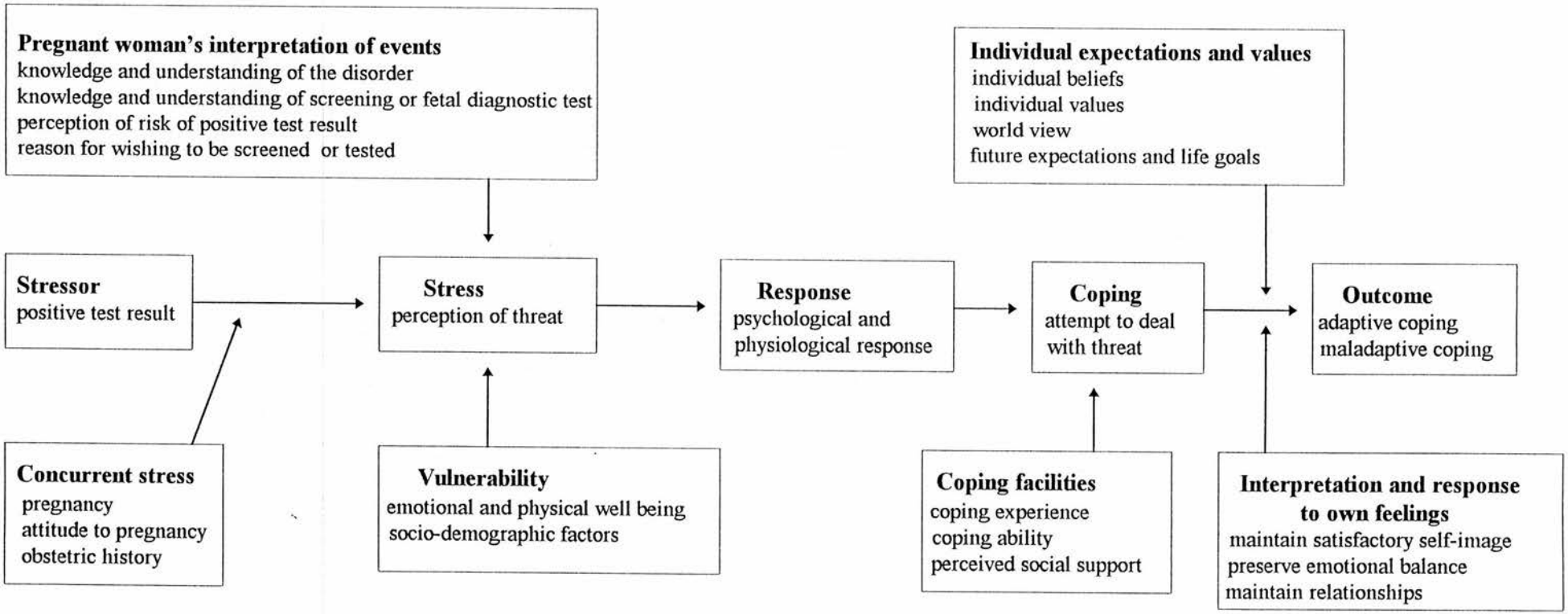


Figure: Variables which contribute to a pregnant woman's emotional response to prenatal screening and diagnosis based on Cochrane's model of stress and coping (Cochrane 1983)

initially identified as the circumstance of receiving a positive screening test result. Since twenty five per cent of all women felt anxious about being screened (figure 6.7) the actual offer of the test itself must be viewed as an immediate stressor to a substantial number of women. Indeed, of those who declined the test 38 per cent were made to feel anxious and only 5 per cent felt reassured at the thought of being screened, compared to 23 per cent and 49 per cent respectively of women who were tested (table 6.5). This finding has implications for midwives in the initial presentation of the test. Individuals face difficulty receiving and incorporating information on both cognitive and emotional levels if anxious (Falek 1984). If a woman is made to feel anxious about the offer of the screening test then she may have difficulty absorbing the information and counselling offered by the midwife. For these women stress intervention should commence before pre-screening information and counselling begins. This means establishing a woman's emotional reaction to the offer of screening and acknowledging and identifying the likely source of her apprehension.

Phipps and Zinn (1986) assessed mood disturbance in women undergoing amniocentesis and compared them to a control group who were not undergoing the procedure. Within both groups they identified two groups: "monitors" (information seekers) and "blunters" (avoiders). It is believed that some individuals seek out threatening information and are more likely to show anxiety and depression (Tunis and Golbus 1991). Among the participants in the CF screening trial no correlation was found between either a woman's socio-economic group or her age and feelings of anxiety toward screening. There may, however, be other variables such as personality, obstetric history or education which contribute to anxiety among a quarter of women screened but further research is needed to establish if this is the case.

Although feelings of anxiety are considered a natural response to any situation perceived as threatening and only become a problem when they become extreme either in duration or intensity (Cochrane 1983), how women cope with stress during pregnancy is a concern. Smoking, alcohol and overeating should be discouraged and acceptable coping strategies employed.

Reasons for wishing to be screened.

Women's written quotations revealed that they not only have diverse and often very personal reasons for accepting or declining the test, but that individual knowledge, understanding, values, expectations, and past experiences are equally germane and disparate. Thus women who are offered an identical test in an identical situation will perceive it very differently. Some are quite clear regarding their expectations:

> *"By having the test I will be able to 1) reassure myself and my partner if the expected baby does not have CF, 2) be given time to consider termination if the baby has CF, 3) prepare myself and my family to cope with a CF baby if termination is not a consideration."*

Others wrestle over their decision because of a lack of understanding:

"I don't know what it means. And I haven't heard of it before."

By asking a woman to formulate their reasons for wishing to be screened, or indeed declining to be screened, can assist the midwife detect if a woman has unrealistic expectations of a screening test and allows her the opportunity to correct misconceptions.

Knowledge and understanding of the disorder

A substantial number of women (85 per cent) had previously heard of CF before they were invited to participate in the screening trial. It may be significant that the major source of their information was television, which as a visual medium, may facilitate a

more integrated perspective of the disease, and in turn may influence uptake of screening. Despite a lack of detailed knowledge many were aware from television that respiratory problems were a main feature of the disease and that chest physiotherapy was pivotal in the care of sufferers. Popular drama programmes on television such as 'London's Burning' and 'Medics' have depicted the physical effects of CF and, in the former drama series, the emotional strain placed upon the CF sufferer's family.

The younger age group 16 to 20 years were significantly less likely than older women to have previously heard of the disease. An observational study of women in labour found that women whose knowledge was scant were not given additional information which would have increased their understanding because they were felt by staff to be less likely to comprehend (Kirkham 1983). Questions are an obvious way to obtain information but formulating questions requires a semblance of knowledge and understanding. There is a danger that those women whose need is greatest may gain inadequate information. Information should be tailored to meet individual needs and ample opportunity offered to ask questions or to admit a lack of understanding.

Some women will rely totally on the information given before and at the booking clinic to formulate both their concept of the disease and the screening test. Around 35 per cent of carriers, controls and partners felt they had not received sufficient information prior to attending the clinic and 23 per cent of carriers felt that on hindsight they had not received enough information about the test at the antenatal clinic (table 16.19).

Vulnerability

Social factors

Women from the extreme ends of the age spectrum were significantly more likely to present at booking with symptoms of stress revealed through the threshold GHQ (figure

6.9). Younger women were significantly less likely to be married (table 6.3) and may lack a supportive relationship. It is recognised that social factors impinge upon coping response (Cochrane 1983) and social support is recognised as a buffer against stress (Cobb 1976). Thirty per cent of all women had not discussed the test with anyone other than the midwife (figure 6.5). This is evidence that a substantial number of women depend upon the midwife for support during the decision making process. Midwives may themselves be unaware of the assignment they undertake and the demand this places upon them, in addition to providing practical care and emotional support to women during the early stages of pregnancy. Midwives will require adequate professional and personal support if they are to take responsibility for prenatal screening counselling and continue providing the high standard of care their work demands.

There is evidence that nurses find caring for individuals' physical needs much less stressful than care which involves more personal and emotional interaction (Menzies 1960). A study carried out in a gynaecological ward revealed that nurses are frequently inexact in their perception of the worries of their patients; worries which fellow patients easily related to (Johnston 1982). An additional concern is that role conflict could arise for the midwife who finds herself wishing to spend longer with a woman who needs her, yet is aware that there may be a backlog of women waiting to be seen.

Younger women were significantly more likely to experience difficulty in comprehending pre-screening information in the leaflet, and over 70 per cent of 16 to 20 year old women and 56 per cent of 21 to 25 year old women either had no perception of their carrier risk, or perceived it wrongly. There was no correlation between lack of knowledge and the decision to decline screening which supports the evidence that some women accept prenatal screening without fully understanding the implications (Marteau et al 1988b). There may be an inherent problem regarding the current mode of delivery

of information by leaflet and discussion. There is undoubtedly scope for research concerned specifically with communication media. Research should seek to evaluate the most suitable and acceptable forms of delivering information, taking into account multiple variables such as age, socio-economic status, educational background, past obstetric history and individual preference. A greater awareness of individual requirements with a wider range of educational aids about prenatal screening is needed.

Education is an important aspect of the midwife's work. Most women are receptive to information in pregnancy because they wish to have a healthy baby. Indeed 26 per cent of women who commented on their decision to be screened, either wished to prevent the birth of an affected child or expressed the wish for a healthy baby. It is important that the midwife explain the main symptoms and variable severity of the disease, along with the possibility for treatment and current life expectancy of those with the disorder. The availability of a leaflet detailing these facts can be a useful adjunct to a pre-screening leaflet, which may offer only a brief outline of the disease. All leaflets should be periodically revised to ensure that new developments in treatment and changes in prognosis are up-to-date. In parallel, in-service education is required to ensure that there are opportunities for midwives to update their knowledge. In a study of midwives knowledge of maternal serum alpha-fetoprotein screening, 45 per cent lacked basic knowledge about the test, even although all were involved in the delivery of screening (Sanden 1985).

Individual counselling requires clear objectives (Jones 1991). With regard to pre-screening counselling the objective is to help a woman and her partner decide whether a test is right for them. In order to structure the counselling process the midwife requires to "listen" to define what a woman needs to know. Counselling requires both

time and space. It requires privacy and freedom from interruption a situation not always possible in a busy antenatal clinic. Does an antenatal booking interview facilitate counselling for genetic screening? This is an area which requires further research, particularly in relation to midwifery staff's own perception regarding their eligibility to deliver genetic screening. Within the antenatal clinic where this study took place, a majority of midwives had attended counselling courses and were trained in how to present information and discuss screening. In addition they had the support of a genetic nurse for more sophisticated information and counselling. Without formal training, delivering prenatal screening could be stressful for midwifery staff and result in women being underinformed or suffering from screening related morbidity as a result of poor preparation for screening. Access to appropriate training in genetics and genetic counselling techniques should be available to staff through the regional genetics service (Modell 1992).

The broad aim of pre-screening information and counselling is to meet the needs of individual women through good communication, provision of adequate information and emotional support. Knowledge is necessary to formulate accurate expectations, make an informed decision and prepare psychologically for screening. Caplan (1964) advises that primary prevention can either alter stressful conditions or strengthen the individual to resist stress and cope in adversity. Primary prevention against a stressful reaction to screening needs to commence during the pre-screening stage with information about the objectives of screening, addressing reasons for wishing to be screened, individual perception of risk, the meaning of a positive test result, the likely course of action and the procedure for reporting a positive test result. Concurrent stress should be identified and steps taken at this time to deal with the cause and alleviate symptoms. In addition women and their partners should be encouraged to assess their own beliefs and values and their perceived ability to cope with a child with a chronic

disease. Their attitude to termination of pregnancy should also be addressed. Adequate pre-screening preparation will influence both primary appraisal and ability to cope with a positive test result by ensuring that women are knowledgeable about the screening process and feel in control.

A framework for pre-screening counselling.

A woman should be given accurate and unbiased information so that an informed decision can be made. Counselling should be non-directive, but should actively help woman reach a decision in the context of her unique social, cultural, personal, family and obstetric situation. A woman has autonomy, that is she has the right to decide what action or inaction she wishes to take. Screening information should be communicated in such a way as to ensure a woman can understand it. Medical jargon should be avoided and opportunity to admit difficulty in understanding should be offered. If a prenatal test is a local screening policy it should be offered to all women. The decision to offer or not to offer a test does not belong to the individual midwife or doctor but to the woman herself. A woman has a right to confidentiality; test results should not be divulged to any other party even if it has implications to other family members. The decision to inform relatives even if the result has implications for them is that of the woman herself.

7.2.2 Care of women who receive a positive CF carrier test result: a basis for coping

When a woman receives a positive screening test result her understanding of her problem will determine her reaction, the way she copes, and the type and amount of counselling and support intervention she requires. Cochrane advises that how an individual deals with the challenge of a stressful event depends on their psychological well being at the time and upon their self-esteem. Belief in one's coping ability is also associated with positive adaptation and well-being (Cochrane 1983). Thus, some women may have a negative outlook about their ability to cope when they receive a positive test result and

need help and encouragement. They may blame themselves, feel guilt or shame and become extremely upset and confused. Alternatively they may blame the doctor or midwife for thrusting this problem on them and become angry or non-communicative. A woman may also be influenced by what she perceives to be an acceptable reaction and hide her true emotions (Cochrane 1983). An awareness of the influences contributing to a diversity of reactions, symptoms and ability to cope are a pre-requisite to providing counselling and support to women faced with a positive test result.

The results of psychological assessment of women and their partners undergoing CF screening in pregnancy concluded that 53 per cent of women experienced a stressful reaction at the time of receiving their positive test result. Information and choices need to be made available to these women and their partners but in conjunction with stress intervention to manage their stress and facilitate coping. Once a favourable outcome was known women identified as carriers regained psychological homeostasis and maintained this for the remainder of their pregnancy (figure 6.13). Carriers and control subjects who were experiencing concurrent stress continued to manifest symptoms of psychological disturbance (table 6.10). This demonstrates a need for adequate follow-up support to ensure emotional adjustment after the screening experience and to monitor prolonged stress.

When a woman receives a positive test result symptoms of stress may be so severe that she has lost sleep, taken time off work or had difficulty functioning as normal. A woman's perception of the threat of her positive screening test result will be influenced by her knowledge, beliefs, expectations, social support, present mood, her confidence in the screening programme and staff and her past ability to cope with stressful situations. From a midwifery care perspective, a woman will be susceptible to change depending upon the influences of the variables outlined in the model.

Moreover, one partner's response can influence the other. Male partners were found to be significantly more likely to manifest a stressful response if their female counterpart was distressed. The midwife too has the capability to influence a woman's perspective of her situation through stress intervention strategies. Within the model a woman may be viewed as someone in a state of flux as she modifies her view of her situation through new information, counselling and her attempts to cope.

Even prior to screening, it is important to gain a broad impression of a woman's current thoughts and feelings about a screening test, her expectations and her belief about her own ability to cope if she received a positive result. The conceptual model can serve as a framework to consider those areas which may influence a woman's response to a positive test result. These include the personal meaning of the test, the likely impact if positive, the psycho-social context in which screening will be carried out, for example her past obstetric history, present pregnancy and life events.

It is clear that a substantial number of women (32% of the screened population in this study) present with signs of psychological disturbance before they are screened. Many (44%) were suffering from early symptoms of pregnancy, others gave a variety of reasons for their distress (Table 6.10). The significance of concurrent stress, in relation to prenatal genetic screening, is that it may amplify the distress of receiving a positive screening test result and the likelihood of inducing sequential stress.

A majority of carriers and partners (80% and 90% respectively) were glad they had been screened. Nonetheless, 20 per cent of carriers either regretted or were ambivalent about having been screened. Although pre-pregnancy screening is arguably the ideal time to offer genetic carrier screening, for many individuals the first real opportunity occurs post-conception.

Psychological preparation has been shown to help patients undergoing a variety of potentially stressful procedures (Ridgeway and Mathews 1982; Bailey and Clarke 1989; Hunter 1994). The midwife is usually the first person that a pregnant woman consults when she attends for prenatal care. The midwife has a crucial role to play in providing unbiased accurate information about the various prenatal screening tests and in guiding them through the procedures which lie ahead. The results of this thesis show that it is not sufficient for the midwife simply to equip women with the facts about a screening test. If the midwife is to deliver screening with an element of caring, psychological preparation of a woman for the unlikely event of a positive test result is important if screening-related morbidity is to be avoided.

Stress interventions to assist patient coping

Midwifery interventions at this stage should involve giving thought to the timing of reporting a positive test result. Choosing a time to divulge the test result when both partners are at home will prevent a situation of a woman coping alone with her feelings. Details of occupation will give some idea of a couple's routine. When the result is divulged primary appraisal of the stressful situation will occur (Cochrane 1983). If adequate pre-screening information and counselling has been carried out it should help a woman feel more in control (Hunter 1994; Marteau and Chapple 1992a; Statham and Green 1993).

By the time a couple are seen for counselling they will have had time to reappraise the challenge of the woman's positive test result (secondary appraisal) and assessed their ability to cope. Any symptoms of stress reaction to the threat will be felt as results of the GHQ and SRT assessments showed (figure 6.13; table 6.13). Stress intervention should deal with a couples emotional response first (Hunter 1994). Hunter advises that

direct coping strategies should be employed. Direct coping strategies involve trying to normalise emotional reactions and reassure that help is at hand to assist coping. Providing a couple with a climate of safety and security by outlining briefly the agenda of the counselling session, the testing procedure, and prevent them from feeling overwhelmed by the situation. By breaking the crisis down into a series of steps can make it feel more manageable (Hunter 1994). Reassurance that the pregnancy is not under threat is an immediate task although false reassurance should be avoided. A realistic comfort is that the CF carrier test has screened the mother not the baby.

Promoting objectivity and accurate secondary appraisal can help. Often the person in crisis has a poor grasp of reality and thinks emotionally and irrationally. However, Caplan (1964) advises that during a crisis the individual is open and amenable to outside intervention. Many couples are unfamiliar with genetic terminology and concepts. Using a visual aid such as a felt board is a non-threatening technique which can help reduce anxiety about something intangible - an altered gene. Issuing a leaflet which reiterates the counselling information is a reassuring additional source of information.

Discouraging loss of self-worth and promoting a positive self view can prevent self-deprecation and diminished well-being (Kessler 1984). Explaining that all individuals carry several altered genes and emphasising the normality of the 'carrier state' is vital in preventing feelings of inadequacy or guilt and answers the question "why me?"

The conceptual model illustrates the ultimate goal of stress intervention is to help the individual strive for effective coping (Cochrane 1983). Focusing upon certain short term goals to increase mastery of the situation by advising a woman how to look after her physical and psychological well being over the period awaiting the partner's test result is important (Hunter 1994). Palliative coping strategies should be addressed not least to

ensure that unsuitable methods such as smoking, drinking drug or exercise abuse are not pursued. Discussing available social support and encouraging the use of the hospital contact phone number to answer questions, concerns and give ongoing emotional support are all positive interventions which aim to achieve the goal of effective coping and reduce stress in the pregnant woman and her partner.

Although the studies in this thesis have concentrated upon the majority of women who are offered a genetic screening test, and on those women who received a positive result but whose partners received a negative test result, there is the small but important group who ultimately face a 1 in 4 risk of having a child with CF. As the advent of team midwifery allows the midwife to devote her time to the care of a small case load of mothers (Department of Health 1993; Scottish Home and Health Department 1993) some midwives will experience the anguish of the woman who is confronted with a high risk of an affected infant. One woman whose partner was also found to be a CF carrier described her experience as follows:

> *"I knew about cystic fibrosis but I didn't know that as many as one in twenty-five people in Britain are carriers - even then I just thought I would be one of the twenty-four. My reaction when I found my husband was also a carrier was one of total disbelief. After everything I had been through, all I could think was why? why? I was heartbroken, I wept buckets."*
> (Bodmer and McKie 1994 page 229-230)

7.3 Final Conclusions

If genetic screening were to become focused on pregnancy it could place women in an unfair position of responsibility for genetic disease in the community. Informed choice is important in antenatal care and choice can only be exercised from a position of

knowledge. To withhold information about a test which is available is wrong but on the other hand women should not feel pressurised into accepting screening. There may be an underlying assumption that genetic screening will develop if normalised into the package of testing offered at booking. Screening should be offered through other channels such as GP surgeries and family planning clinics. An alternative method of screening is to test both partners of a couple, thereby avoiding the focus of attention to be solely on the woman (Chapple 1992).

Most women want prenatal screening. Although there are practical, emotional and moral reasons for not using it, it does seem that most women feel the need to know the knowable (Rothman 1988). Many want information about their fetus because they want to plan. The nursing and midwifery professions need to plan also for the advent of genetic screening. Currently there is an imbalance between the rapid advance of knowledge and practice in the research laboratory and the lack of knowledge and practice at the clinical level, where both benefit and harm to the individual can occur. The need for counselling and emotional support, along with detailed educational materials as an integral part of genetic screening has implications for both nurse managers and nurse educationalists. At the managerial level the cost of offering a screening programme poses a serious financial decision.

Prenatal screening involves laboratory costs, staff time and education, patient information and support services of prenatal diagnosis and termination of pregnancy. In addition service monitoring must be organised and this requires clerical assistance and finance. Thus the decision as to whether to fund new developments such as CF carrier screening may create a problem. There is also a danger that in a stringent financial climate prenatal CF carrier screening programmes may be initiated without proper education of staff and a counselling framework. For nurse educationalists and managers there is the responsibility

of ensuring that staff have the necessary knowledge and training to present the complex issues of genetic screening. Most allegations of negligence in the field of prenatal diagnosis, rather than related to misdiagnosis or fetal loss, have revolved around failure to screen, misinformation, risk assessment or counselling. Few examples have been published as a result of out of court settlements (Modell 1992).

As midwives enter into a new era of midwife-led care their responsibilities will extend across the whole domain of antenatal management. If pregnancy is to be used as one gateway to genetic screening then there is a pressing need for midwives to develop skills in counselling and to know how to co-ordinate the care of women with obstetric-genetic needs with experts in genetic counselling The concept of women-centred care ties in well with the ethos of genetic counsellors. Indeed continuity of care in midwifery will lend itself particularly well to the needs of women who undergo genetic screening. Ideally the same counsellor should be available to help women reach the right decision for them in the context of their unique social, moral and medical situation (Modell 1992).

Although it seems unlikely that genetic screening causes harm to women who test negative there may be subtle impairment in the form of overloading of antenatal information. As the number of prenatal screening tests increase the risk of information overload, blocking of information and misinterpretation of other antenatal health care messages could foreseeably occur. This could affect a woman's attitude both to pregnancy and to the midwife. Further research is needed to clarify these uncertainties.

As molecular biology increases knowledge about the cause of genetic diseases and develops ways of preventing them, so too will it continue to try to remedy the effects through gene therapy. Increasingly pre-screening counselling will involve providing information not only about preventive strategies but about therapies. However, most

pregnancies do not result in fetal abnormality but in the birth of a healthy infant and herein may lie one of the most important roles of the midwife in the area of prenatal screening: safeguarding the balance of antenatal information so that discussion of abnormalities of pregnancy is not allowed to outweigh description of the normal.

A limitation of the research carried out in this thesis is that it fails to address the question of midwives' attitudes to genetic screening per se; to the offer of a genetic screening test to pregnant women; and more specifically how midwives view their taking the responsibility for presenting genetic screening. There is a need for these questions to be answered before further genetic screening programmes are initiated.

REFERENCES

Abbott P, Sapsford R. (Eds) (1993) Research into Practice. Open University Press

Abuelo DN, Hopmann MR, Barsel-Bowers G, Goldstein A. (1991) Anxiety in women with low maternal serum alpha-fetoprotein screening results. Prenat Diagn 11: 381-385.

Adler B, Kushnick T. (1982) Genetic counselling in prenatally diagnosed Trisomy 18 and 21: Psychosocial aspects. Pediatrics 69: 94-99.

Aguilera DC. (1990) Crisis intervention. Theory and methodology (6th edition). The C.V. Mosby Company, USA.

Allen I. (1987) Education in Sex and Personal Relationships. Frames Pinter Ltd. London.

Andersen DH. (1938) Cystic fibrosis of the pancreas and its relation to celiac disease: A clinical and pathological study. Am J Dis Child 56:344-399.

Anderson WF. (1992) Human Gene Therapy. Science 256: 808-813.

Andeson-Segesten K, Plos K. (1988) The needs, concerns and coping of mothers of children with cystic fibrosis. Scand J Caring Sci 3: 35-41.

Andrews B, Brown GW. (1988) Marital violence in the community: a biographical approach. Br J Psychiat 153: 305-319.

Angastiniotis M. (1990) "Cyprus" Thalassaemia programme." Lancet 336: 119-1120.

Anionwu E. (1991) Genetics: a multi-ethnic approach. Nursing 4 No 41: 9-12.

Anonymous. (1989) When risk factors have little meaning. BMJ 299; 6715: 1599-1600.

Antley RM. (1976) Variables in the outcome of genetic counselling. Soc Biol 23: 108-115.

Antley RM, Hartlage LC. (1973) The effects of genetic counselling for Down's syndrome. Paed Res 7: 345.

Astbury J, Walters WAW. (1979) Amniocentesis in the early second trimester of pregnancy and maternal anxiety. Australian Family Physician 8: 595-599.

Bailey R, Clarke M (1989) Stress and Coping in Nursing. Chapman Hall, London.

Baird PA. (1990) Opportunity and danger: medical, ethical and social implications of early DNA screening for identification of genetic risk of common adult onset disorders. In: Knoppers BM; Laberge CM. (Eds) Genetic screening: from newborns to DNA typing. Excerpta Medica, New York.

Banks MH (1983) Validation of the General Health Questionnaire in a young community sample. Psychol Med 13: 340-353.

Barker PJ (1991) Questionnaire. In: Cormack PJ (ed) The Research Process in Nursing 2nd edition Blackwell Scientific Publications, Oxford chapter 23.

Basford P. (1990) How to use questionnaires. Nursing Times 86;49: 57.

Beaudet AL. (1989) Cystic fibrosis. In: Scriver CR, Beaudet AL, Sly WS, Valle D. Eds. The metabolic basis of inherited disorders. 6th ed. New York: McGraw-Hill: 2649-2680.

Bebbington P, Hurry J, Tennant C, Sturt E, Wing J. (1981) Epidemiology of mental disorders in Camberwell. Psychol Med 11: 561-581.

Beeson D, Golbus MS. (1979) Anxiety engendered by amniocentesis.Birth Defects Original Article Series 15: 191-197.

Bell J. (1990) Prenatal diagnosis: current status and future trends. In: Human Genetics Information: Science, Law and Ethics. Ciba Foundation Symposium 149. John Wiley and Sons, Chichester, UK

Bennett MJ, Gall GS, Gau DW. (1980) Women's attitudes to screening for neural tube defects. Br J Obstet Gynaecol 87: 370-371.

Berkowitz G, Kasl S. (1983) The role of psychosocial factors in spontaneous preterm delivery. J Psychosom Research 27: 283-290.

Berne -Fromell K, Kjessler B, Josefson G. (1983a) Anxiety concerning fetal malformations in women who accept or refuse alpha-fetoprotein screening in pregnancy. J Psychsom Obste Gynaecol 2:94-97.

Berne-Fromell K, Uddneberg N, Kjessler B. (1983b). Psychological reactions experienced by pregnant women with an elevated SAFP level. J Psychosom Obstet Gynaecol 2:233

Berne-Fromell K, Kjessler B. (1984) Anxiety concerning fetal malformations in pregnant women exposed or not exposed to an antenatal serum alpha-fetoprotein screening programme. Gynecol Obstet Invest 17: 36-39.

Berryman JC, Windridge K. (1991) Having a baby after 40: Part I: A preliminary investigation of women's experience of pregnancy. J Reprod Infant Psychol 9: 3-18.

Bewley CA, Gibbs A. (1991) Violence in pregnancy. Midwifery 7: 107-112.

Black RB, Furlong R. (1984) Prenatal diagnosis: the experience in families who have children. Am J Med Genet 19: 729-739.

Bluglass K. (1984) Early infant loss and multiple congenital abnormalities. In: Emery AEH, Pullen I. Psychological aspects of genetic counselling. Academic Press, London.

Blum BL. (1980) Psychological aspects of pregnancy: birthing and bonding. Human Sciences Press, New York.

Blumberg BD, Golbus MS, Hanson KH. (1975) The psychological sequelae of abortion performed for genetic indication. Am J Obstet Gynecol 122: 799-808.

Blumberg B. (1984) The emotional implications of prenatal diagnosis. In: Emery AEH, Pullen I. (Eds) Psychological Aspects of Genetic Counselling. Academic Press, London.

Boat TF, Welsh MJ, Beaudet AL. (1989) Cystic Fibrosis In: Scriver CR, Beaudet AL, Sly WS, Valle D. (Eds) The metabolic basis of inherited disorders. 6th ed. New York: McGraw-Hill: 2649-2680.

Bodmer W, McKie R. (1994) The Slippery Slope ch 13 in The Book of Man. Little, Brown and Company UK.

Bohn DK. (1990) Domestic violence and pregnancy. Implications for practice. J Nurse Midwifery: 35(2): 86-98.

Bothamley J. (1990) Are fathers getting a fair deal? (in childbirth and parenting). Nurs Times 88(36): 68-69.

Bourne S, Lewis E. (1984) Pregnancy after stillbirth or neonatal death: psychological risks and management. Lancet ii: 31-33.

Briscoe M. (1989) The detection of emotional disorders in the post-natal period by health visitors. Health Visitor 62: 336-338.

Brock DJH, Sutcliffe RG. (1972) Alphafetoprotein in the antenatal diagnosis of anencephaly and spina bifida. Lancet ii 197-199.

Brock DJ (1984) Cystic Fibrosis. In: Wald NJ (ed) Antenatal and Neonatal Screening. Oxford University Press, New York.

Brock DJH. (1990a) Population screening for cystic fibrosis. Am J Hum Genet 47: 164-165.

Brock DJH. (1990b) Heterozygote screening for cystic fibrosis. Unpublished research proposal submitted to the The Cystic Fibrosis Trust.

Brock DJH, Mennie ME, McIntosh I, Jones CE, Shrimpton AE. (1991) Heterozygote screening for cystic fibrosis. In: Drife JO and Donnai D (Eds) Antental diagnosis of fetal abnormalities. Springer-Verlag, London.

Brock (1993) Prenatal diagnosis of cystic fibrosis. In:Dodge JA, Brock DJH, Widdicombe JH. (Eds) Cystic fibrosis: current topics. John Wiley and Sons page 63.

Brockington IF, MartinC, Brown GW, Goldberg D, Margison F. (1990) Stress and Peurperal Psychosis. Br J Psychiat 157:331-334.

Brown MA. (1986) Marital support during pregnancy. JOGN November/December: 475-483

Brown GW, Andrews BA, Harris TO, Adler Z, Bridge L. (1986) Social support, self esteem and depression. Psychol Med 16:813-831

Brown GW, Adler Z, Bifulco A. (1988) Life events, difficulties and recovery from chronic depression. Br J Psychiat 152: 487-498.

Brown GW, Lemyre L, Bifulco A. (1992) Social factors and recovery from anxiety and depressive disorders. A test of specificity. Br J Psychiat 161: 44-54.

Bryman A (1990) Quantity and Quality in Social Research. 2nd edit Unwin Hyman, London.

Bull MJV (1990) Maternal and fetal screening for antenatal care. BMJ 300: 1118-1120.

Burn J (1993) Screening for cystic fibrosis in primary care. Family practice at last? BMJ 306: 1558-1559.

Burton BK, Dillard RG, Clark EN. (1985a) The psychological impact of false positive elevations of maternal serum alpha-fetoprotein. Am J Obstet Gynecol 151: 77-82.

Burton BK, Dillard RG, Clark EN. (1985b) Maternal serum alpha-fetoprotein screening: the effect of participation on anxiety and attitude toward pregnancy in women with normal results. Am J Obstet Gynecol 152: 540-543.

Bury J. (1984) Teenage Pregnancy in Britain. Birth Control Trust, London.

Campbell J. (1986) Nursing assessment for risk of homicide with battered women. Adv Nurs Sci 8(4): 36-51.

Campbell IE, Field PA. (1989) Common psychological concerns experienced by parents during pregnancy. Canada's Mental Health 37: 2-5.

Cao A, Rosatelli C, Galanello R. (1989) The prevention of thalassaemia in Sardinia. Clin Genet 36: 277-285.

Caplan, G. (1964) Principles of preventive psychiatry. Basic Books, New York. Cited in: Ewing, C.P. (1978) Crisis intervention as psychotherapy. Oxford University Press, New York.

Cappelli M, McGrath PJ, MacDonald NE. (1989) Parenatal care and over-protection of children with cystic fibrosis. Br J Med Psychol 62: 281-289.

Carrell S. (1992) New hope in fighting killer lung disease. The Scotsman 24th July page 3.

Carty EM, Conine TA, Hall L. (1990) Comprehensive health promotion for the pregnant woman who is disabled. J Nurs Midwifery 35(3): 133-142

Chan V, Chan TK, Todd D. (1991) Thalassaemia screening and prenatal diagnosis in Hong Kong. Am J Hum Genet 49(Supp): 79.

Chapple J, Dale R, Evans BG. (1987) The new genetics: will it pay its way? Lancet **i:1189-**1192

Chapple JC. (1992) Genetic Screening. In: Brock DJH, Rodeck CH, and Ferguson-Smith MA. (Eds) Prenatal Diagnosis and Screening. Churchill Livingstone, Edinburgh. page 579-593.

Chevrin A, Farnsworth PB, Freedman WL, Duncan PA, Shapiro LR. (1977). Amniocentesis for prenatal diagnosis. N.Y. State J Med August 1

Childs B, Gordis L, Kaback MM. (1976) Tay Sachs screening: social and psychological impact. Am J Hum Genet 28: 550-558.

Christie B. (1992) Screen trial offers hope of genetic disease cut. Scotsman October 23rd page 6.

Clark SL, DeVore GR. (1989) Prenatal diagnosis for couples who would not consider abortion. Obstet Gynecol 73: 1035-1037.

Clarke A. (1993) Ethical issues raised by the new genetics. In: Young I.D. (Ed) The New Genetics. Clinical Paediatrics. Bailliere Tindall, London.

Clarke JTR, Skomorowski MA, Zuker S. (1989) Tay-Sachs disease carrier screening: follow-up of a case finding approach. Am J Med Genet 34: 601-605.

Clinton JF. (1987) Physical and emotional responses of expectant fathers throughout pregnancy and the early postpartum period. Int J Nurs Stud 24(1): 59-68.

Clothier C. (1992) Report of the committee on the ethics of gene therapy. London HMSO.

Clow CL, Scriver CR. (1977) Knowledge about and attitudes toward genetic screening among high-school students: the Tay-Sachs experience. Pediatrics 59: 86-91.

Cobb E, Holloway S, Elton R, Raeburn JA. (1991). What do young people think about screening for cystic fibrosis? J Med Genet 28: 322-324.

Cobb S. (1976) Social support as a moderator of life stress. Psychosom Med 38: 300-314.

Cochrane RA. (1980) A comparative evaluation of the Symptom Rating Test and the Langer 22-item Index for use in epidemiological surveys. Psychol Med 10: 115-124.

Cochrane R, Stopes-Roe M. (1981) Women, marriage, employment and mental health. Br J Psychiat 139: 373-381.

Cochrane R. (1983) The Social Creation of Mental Illness. Longman, London.

Cohen FL. (1984) Clinical genetics in nursing practice. Philadelphia: Lippincott chapter 9: page 156.

Cohen LF, di Sant'Agnese PA, Friedlander J. (1980) Cystic fibrosis and pregnancy: A national survey. Lancet 2: 842-844.

Colten HR. (1990) Screening for cystic fibrosis. Public policy and personal choices. N Eng J Med 332(5): 328-329.

Condon JT. (1987) Psychological and physical symptoms during pregnancy: A comparison of male and female expectant parents. J Reprod Infant Psychol 5: 207-213.

Cookson C. (1992) The men who would play God. Weekend Financial Times February 1/2

Cooper CL, Marshall J. (1978) Understanding Executive Stress. Macmillan, London. Cited in: Hingley P, Cooper C. (1986) Stress and the Nurse Manager. John Wiley and Sons, Chichester.

Coutelle C. (1993) Gene therapy for cystic fibrosis. Arch Dis Child 68(4):437-440

Cox T. (1978) Stress. Macmillan, London.

Cox JL, Connor Y, Kendell RE. (1982) Prospective study of psychiatric disorders of childbirth. Br J Psychiat 140: 111-117.

Crawfurd M. (1983) Ethical and legal aspects. Br Med Bull 39(4): 310-314.

Crespigny de L, Dredge R. (1991) Which tests for my unborn baby. Oxford University Press, Australia

Crossfield T. (1988) How to devise a questionnaire. Nursing Standard 3 (5): 39.

Crossley JR, Elliott RB, Smith PA. (1979) Dried blood spot screening for cystic fibrosis in the newborn. Lancet 1: 472.

Crow MM. (1972) "True to Life" : a relevant approach to patient education. Am J Public Health 62: 1328-1330.

Cuckle HS, Wald NK, Cuckle PM. (1989) Prenatal screening and diagnosis of neural tube defects in England and Wales in 1985. Prenatal Diagn 9: 393-400.

Cuckle HS. (1990) Screening with discrimination. Arguments for a national screening body. BMJ 301: 889-890.

Cuckle HS, Wald NJ. (1984) Principles of screening. In: Wald NJ (Ed) Antenatal and neonatal screening. Oxford University Press, Oxford page 1-21.

Cuckle HS. (1992) Down's syndrome screening: background publications. Institute of Epidemiology 34, Hyde Terrace, Leeds.

Cummings MR. (1988) Human Heredity, Principles and Issues. West Publishing, St. Paul.

Cunningham GC, Gilstrap LC. (1991) Maternal serum alpha-fetoprotein screening N Eng J Med 325: 55-57.

Curry M (1994) Antenatal tests:reassuring or stress provoking? Professional Midwife August 1994 21-23.

Davies BL, Doran TA. (1981) Factors in a woman's decision to undergo genetic amniocentesis for advanced maternal age. Nurs Res 31 (1) 56-59.

Davies K, Williamson R. (1993) Gene therapy begins. BMJ 306:1625-1626.

Decruyenaere M, Evers-Kiebooms G, Denayer L, Van den Berghe H. (1992). Cystic fibrosis: community knowledge and attitudes towards carrier screening and prenatal diagnosis. Clin Genet 41: 189-196.

Department of Health (1993) Changing Childbirth. Report of the Expert Maternity Group Part 1. HMSO, London.

Dodge JA, Morison S, Lewis PA, Coles EC, Geddes D, Russell G, Jackson AD, Bentley B. (1993). Cystic fibrosis in the United Kingdom 1968-1988: incidence, population and survival. Paed Perin Epidem 7: 157-166.

Donnai P, Charles N, Harris R. (1981) Attitudes of patients after genetic termination of pregnancy. BMJ 282: 621-622.

Donnai D. (1992) Genetic counselling and the the pre-pregnancy clinic. In: Brock DJH, Rodeck CH, Ferguson-Smith MA. (Eds) Prenatal Diagnosis and Screening. Churchill Livingstone, Edinburgh. page 3-10.

Drake ML, Verhulst D, Fawcett J. (1988) Physical and psychological symptoms experienced by Canadian women and their husbands during pregnancy and the postpartum. J Adv Nurs 13:436-440.

Duffy ME (1985) Designing nursing research: the qualitative / quantitative debate. J Adv Nursing 10: 225-232.

Duncan-Skingle F, Foster F. (1991) The management of cystic fibrosis. Nurs Stand 5(21): 32-34

Ekwo EE, Kim J, Gosselink CA. (1987) Parental perceptions of the burden of genetic disease. Am J Med Genet 28: 955-963.

Elborn JS. (1991) Cystic fibrosis: Screening or cure? Ethics and Medicine 7(3): 40-42.

Elias S, Annas G, Simpson JL. (1991) Carrier screening for cystic fibrosis: implications for obstetric and gynecologic practice. Am J Obstet Gynecol 164: 1077-1083.

Elkins J, Stovall TG, Wilroy S, Davis JV. (1986) Attitudes of mothers of children with Down's syndrome concerning amniocentesis, abortion and prenatal genetic counselling techniques. Obstet and Gynecol 68: 181-184.

Elliott BJ, Huppert FA. (1991) In sickness and in health: associations between physical and mental well-being, employment and parental status in a British nationwide sample of married women. Psychol Med 21(2): 515-524.

Elliot SA, Rugg Aj, Watson JP, Brough DI. (1983) Mood changes during pregnancy and after the birth of a child. Br J Clin Psychol 22: 295-308.

Ellis I. (1991) Carrier screening for Tay-Sachs. Nursing 4 No 41: 16-18.

Emery AEH. (1984) The principles of genetic counselling. In: Emery AEH, Pullen I. (Eds) Psychological Aspects of Genetic Counselling. Academic Press. London.

Emery AEH, Pullen I. (Eds) (1984) Psychological Aspects of Genetic Counselling. Academic Press, London.

Emery AEH, Raeburn JA, Skinner R, Holloway S, Lewis P. (1979) Prospective study of genetic counselling. BMJ 1:1253-1256.

Faden RR, Chwalow AJ, Orel-Crosby E, Holtzman NA, Chase GA, Leonard CO. (1985) What participants understand about a maternal serum alpha-fetoprotein screening program. Am J Public Health 75: 1381-1384.

Faden RR, Chwalow J, Quaid J, Chase GA. (1987) Prenatal screening and pregnant women's attitudes towards the abortion of defective fetuses. Am J Pub Health 77(3):288-290

Falek A. (1984) Sequential aspects of coping and other issues in decision making in genetic counselling. Emery AEH, Pullen I. (Eds). Psychological aspects of genetic counselling. Academic Press, London.

Family Circle (1991) Cystic fibrosis. "The test every parent should have." September 1992 , IPC Magazines Ltd. King's Reach Tower, Stamford Street, London.

Farrant W. (1985) Who's for amniocentesis? The politics of prenatal screening. In: Homans H (Ed) The sexual politics of reproduction. Gower, London.

Farrant W. (1980) Stress after amniocenteis for high serum alpha-fetoprotein concentrations. BMJ 281: 452.

Fava GA, Kellner R, Michelacci L, Trombini G, Pathak D, Orlandi C, Bocicelli L. (1982) Psychological reactions to amniocentesis: a controlled study. Am J Obstet Gynecol 143: 509-513.

Fava GA, Kellner R, Perini GI, Fava M, Michelacci L, Munari F, Evangelisti LP, Grandi S, Bernardi M, Mastrogiacomo I. (1983) Italian validation of the Symptom Rating Test (SRT) and Symptom Questionnaire (SQ) Can J Psychiat 28: 117-123.

Fearn J, Hibbard BM, Laurence KM, Roberts A, Robinson JO. (1982) Screening for neural tube defects and maternal anxiety. Br J Obstet Gynaecol 89: 218-221.

Ferrie RM, Robertson NH, Vaudin S, Schwarz M, Little S. (1991) Development of multiplex ARMS test for the simultaneous detection of four common CF mutations. Am J Hum Genet 49: 993A.

Field PA. (1990) Effectiveness and efficacy of antenatal care. Midwifery 6: 215-223.

Field T, Garcia R. (1985) Effects of ultrasound feedback on pregnancy anxiety, fetal activity and neonatal outcome. Obst Gynecol 66(4): 525-528.

Field PA, Morse JM. (1990) Nursing research: the application of qualitative approaches. London: Chapman and Hall pages 102-103.

Fielding NG, Lee RM. (1991) Using Computers in Qualitative research. Sage Publications, London.

Fisher L, Rowley PT, Lipkin M. (1981) Genetic counselling for β-thalassaemia trait following health screening in a health maintenance organization: comparson of programmed and conventional counseling. Am J Hum Genet 33:987-994.

Fletcher JC, Wertz DC. (1992) Ethics and prenatal diagnosis: past, present and future. In: Brock DJH, Rodeck CA, Ferguson-Smith MA. (Eds) Prenatal Diagnosis and Screening. Churchill Livingstone, Edinburgh.

Fraser FC, Levy EP. (1972) Follow-up of a genetic counselling programme. Am J Hum Genet 24: 30a.

Fraser FC. (1974) Genetic counseling. Am J Hum Genet 26: 636-659.

Freda MC, Anderson HF, Damus K, Merkatz IR. (1993) What pregnant women want to know: a comparison of client and provider perceptions. JOGNN 22 (3): 237-244.

Frets PG, Duivenvoorden HJ, Verhage F, Peters-Romeyn MT, Niermeijer MF. (1991) Analysis of problems in making the reproductive decision after genetic counseling. J Med Genet 28:194-200.

Gage LW, Leidy NK. (1991) Screening for psychosocial distress: implications for prevention and health promotion. Public Health Nursing 8(4): 267-275.

Genetic Nurses and Social Workers Association (1994). Unpublished membership list.Secretary: Christine Patch, Wessex Clinical Genetics Service, Princess Ann Hospital, Coxford Road, Southampton.

Gelehrter TD, Collins FS. (1990) Principles of Medical Genetics Williams and Wilkins, Baltimore.

George MR. (1990) CF not just a paediatric problem any more. R.N. September page 60-64.

Gerzi S, Berman E. (1981) Emotional reactions of expectant fathers to their wives first pregnancy. Br J Med Psych 54: 259-265.

Gibson LE, Cooke RE. (1959) A test for concentration of electrolytes in sweat in cystic fibrosis of the pancreas utilising Pilocarpine by Iontophoresis. Pediatrics 23: 545-549.

Gill M, Murday V, Slack J (1987) An economic appraisal of screening for Down's syndrome in pregnancy using maternal age and serum alphafetoprotein concentration. Social Science and Medicine 24:725-731.

Goffman E. (1963) Stigma: Notes on the Management of Spoiled Identity. Prentice Hall, Englewood Cliffs. NJ.

Golbus MS (1992) Prenatal diagnosis availability (editorial) Am J Med Genet 42:800.

Goldberg D, Hillier VF. (1979) A scaled version of the General Health Questionnaire. Psychol Med 9: 139-145.

Goldberg D, Williams P. (1988) A user's guide to the General Health Questionnaire. NFER-Nelson, Windsor.

Goldberg D, Bridges K, Cook D, Evans B, Grayson D. (1990) The influence of social factors on common mental disorders: destabilisation and restitution. Br J Psychiat 156: 704-713.

Goldberg D, Huxley P. (1992) Common Mental Disorders. A Biosocial Model. Tavistock/Routledge, London.

Goldberg DJ, Johnstone FD. (1993) Universal named testing of pregnant women for HIV Br Med J 306:1144-1145.

Goldstein H, Philip J (1990) A cost-benefit analysis of prenatal diagnosis by amniocentesis in Denmark. Clin Genet 37:241-263

Gombeski Jr WR. (1981) Health information sources of the poorly informed: implications for health educators and communicators. Health Values: Achieving high level wellness 5: 199-200.

Goodchild MC, Dodge JA. (1985) Cystic Fibrosis. Manual of Diagnosis and Management. (2nd edition) Bailliere Tindall.

Goodmillow K, Milano CT, Hirschorn K. (1978) Patient response to genetic prenatal diagnosis - a controlled study. Am J Hum Genet 30: 52A.

Gottesman MM. (1992) Maternal adaptation during pregnancy among adult early, middle and late childbearers: similarities and differences. Mat-Child Nurs J 20(2):93-110.

Green JM. (1990a) 'Who is unhappy after childbirth?' Antenatal and intrapartum correlates from a prospective study. J Reprod Infant Psychol 8:part 3.

Green JM. (1990b) Calming or harming? a critical review of psychological effects of fetal diagnosis on pregnant women. Galton Institute, Occasional Papers, Second Series No 2.

Green J. (1990c) Prenatal screening and diagnosis: some psychological and social issues. Br J Obstet Gynaecol 97 (12):1074-1076.

Green JM. (1990d) Is the baby alright and other worries. J Reprod Infant Psychol 8:225-226.

Green JM. (1992). Principles and practicalities of carrier screening: attitudes of recent parents. J Med Genet. 29:313-319.

Guilbert P. (1988) The role of the nurse in genetic counselling services. Unpublished directive: Genetic Nurses and Social Workers Association, U.K.

Guilbert P, Cheater F. (1990) Health visitors' awareness and perception of clinical genetic services. J Med Genet 27:508-511.

Guthrie R. (1968) Screening for inborn errors of metabolism in the newborn period - a multiple test programme. Birth Defects 4:92-98.

Haan EA. (1990) Ethics and the new genetics. J Paed Child Health 26: 177-179.

Harper PS (1984) Practical Genetic Counselling. John Wright & Sons, Bristol. page 161.

Health Education Board for Scotland (1993) Antenatal care and antenatal classes. In: New Pregnancy Book. Health Education Authority, Hamilton House, Mabledon Place, London.

Hecht F, Holmes L. (1972) What we don't know about genetic counselling. N Eng J Med 287:464-465.

Henderson JG. (1982) Value of a single night-time dose of potassium clorazapate in anxiety: a controlled trial comparison with diazepam. Scott Med J 27:292-296.

Hepper PG. (1989) Foetal Learning:Implications for psychiatry? Br J Psychiat 155: 289-293.

Hockey L. (1991) The nature and purpose of research. In: Cormack D. (Ed) The Research Process in Nursing. Second Edition. Blackwell Scientific Publications, Oxford.

Hockey L. (1976) Women in Nursing. Hodder and Stoughton, London.

Hillard PJA. (1985) Physical abuse in pregnancy. Obstet Gynecol 66:185-190.

Ho E. (1985) Anorexia nervosa in pregnancy. Nurs Mirror 160(17): 40-42.

Hodgkin P, Yoxen E. (1985) Biotechnology and general practice. 2. Beyond the technology - social and ethical problems. J Roy Coll Gen Pract 35: 527-531.

Hodson ME. (1993) Cystic fibrosis. Update 15th March page 451-460

Holland WW, Stewart S. (1990) Screening in Health care. Benefit or Bane? The Nuffield Provincial Hospitals Trust.

Hopper E. (1991) Shattered dreams (counselling bereaved parents after stillbirth / miscarriage) Nurs Stand 6(4): 20-21.

Houston MJR, Field PA. (1988) Practices and policies in the initiation of breast feeding. JOGN 17:418-424.

Hunter M (1994) Counselling in Obstetrics and Gynaecology. Communication and Counselling in Health Care Series.BPS Books (The British Psychological Society) Leicester.

Hyde SC, Gill DR, Higgins CF, Trezise AEO, MacVinish LJ, Cuthbert AW. (1993) Correction of the ion transference defect in cystic fibrosis transgenic mice by gene therapy. Nature 362:250-255

Iles S. (1989) The loss of early pregnancy. Clin Obstet Gynaecol 127: 850-854.

Jiminez MH, Newton N. (1979) Activity and work during pregnancy and the postpartum period: a cross-cultural study of 202 societies. Am J Obstet Gynecol 135: 171-176.

Johnston AW (1990) teaching of medical genetics in Britain: A report from the Royal College of Physicians, London. J Med Genet 27:707-709.

Johnston M. (1982) Recognition of patients' worries by nurses and other patients. Br J Clin psychol 21:255-261.

Johnstone F, Hamilton B, Gore SM. (1992) Socioeconomic factors associated with HIV infection in pregnant women. Hlth Bull 50(2):156-162.

Jones A. (1991). The path towards a common goal Structuring the counselling process. Professional Nurse: March pages 302-306.

Jones K. (1990) Expectant fears. Nurs Times 86(15): 36-38.

Jordan P. (1990) Labouring for relevance: expectant and new fatherhood. Nurs Research 39(1): 11-16.

Jorgensen C, Uddenberg N, Ursing Z. (1985) Ultrasound diagnosis of fetal malformation in the second trimester: the psychological reactions of women. J Psychosom Obstet Gynecol 4:32-40.

Kaback MM, Zeiger RS, Reynolds LW. (1974) Approaches to the control and prevention of Tay-Sachs disease. Progress in Medical Genetics 10:103-134.

Kaplan F. et al (1992) Cystic fibrosis carrier detection by DNA test: a population study: carrier frequencies and attitude toward a test with imperfect sensitivity. Am J Hum Genet 51:A268 (Supplement).

Kellner R, Sheffield BF. (1973) A self-rating scale of distress. Psychol Med 3:88-100

Kellner R, Buckman MT, Fava GA, Pathak D. (1984) Hyperprolactinemia, distress and hostility. Am J Psychiat 141(6):759-763

Kelly PT. (1977) Dealing with Dilemma. Springer-Verlag, New York.

Kelsall J. (1992) "She can lip-read, she'll be all right": improving maternity care for the deaf and hearing impaired. Midwifery 8:178-183.

Kenen RH, Schmidt RM. (1978) Stigmatization of carrier status: social implications of heterozygote genetic screening programmes. Am J Public Health 68:116-1120.

Kessler S, Kessler H, Ward P. (1984) Psychological aspects of genetic counselling. III. Management of guilt and shame. Am J Med Genet 17: 673-679.

Kings Fund Forum (1987) Consensus statement screening for fetal and genetic abnormality. BMJ 295: 1551-1553.

Kirkham MJ. (1983) Labouring in the dark. In: Wilson-Barnett J (Ed).Nursing Research: ten studies in patient care. John Wiley and Sons.

Kitzinger S. (1982) Birth over thirty. Sheldon Press, London.

Kitzinger S. (1987) Freedom and Choice in Childbirth. Viking Press, London.

Kumar R, Robinson K. (1984) A prospective study of emotional disorders in childbearing women. Br J Psychiat 152: 799-806.

Kyle D, Cummins C, Evans S. (1988) Factors affecting the uptake of screening for neural tube defects. Br J Obstet Gynaecol 95:560-564.

Laryea M. (1991) Counselling the pregnant cocaine user. Midw HV Comm Nurse 27(2): 48-50.

Lawson A. (1990) Teenage pregnancy in context: The USA and UK. J Reprod Infant Psychol 8: 228-229.

Lazarus R. (1966) Psychological stress and the coping process. McGraw-Hill, New York.

Lazurus R. (1982). Stress and coping as factors in health and illness. In: Cohen J, Cullen J, Martin LR. (Eds) Psychosocial aspects of cancer. Raven Press, New York.

Lederman RP. (1984) Acceptance of pregnancy. In: Psychosocial Adaptation in Pregnancy. Prentice Hall Inc. New Jersey.

Lennon C. (1989) The structural contexts of stress. J Hlth Soc Behav 30: 261-268.

Leonard CO, Chase GA, Childs B. (1972) Genetic counselling: A consumer's view. New Eng J Med 287: 422-439.

Leschot NJ, Verjaal M, Treffers PE. (1982) Therapeutic abortion on genetic grounds - detailed follow-up study of 20 patients. J Psychosom Obstet Gynecol 1-2: 47-56.

Lester C, Farrow S. (1988) Unplanned pregnancies at antenatal clinic. Midwifery 4: 184-189.

Levine JA. (1991) Are patients in favour of general health screening? J R Soc Med 84: 280-283.

Lewis C. (1986) Becoming a father. Milton Keynes: Open University Press.

Lewiston NJ. (1990) The CF young adult comes of age. Chest 97: 1282-1283

Ley P. (1988) Communicating with patients. Chapman Hall, London.

Lilford RJ. (1991) Invasive diagnostic procedures in the first trimester. In: Drife JO, Donnai D. (Eds) Antenatal Diagnosis of Fetal Abnormalities. Springer-Verlag, London.

Lipkin M, Fisher L, Rowley PT. (1986) Genetic counselling of asymptomatic carriers in a primary care setting. Ann Int Med 105: 115-123.

Lipmann-Hand A, Fraser FC. (1978) Genetic counselling: parents' responses to uncertainty In: Epstein CJR, Packman S, Sherman S, Hall BD (Eds) Risk, communication and decision making in genetic counseling. Alan R Liss for the National Foundation-March of Dimes. BD:OAS XV (5c):355-367.

Lloyd J, Laurence KM. (1985) Sequelae and support after termination of pregnancy for fetal malformation. BMJ 290: 907-909.

Lorenz RP, Botti JJ, Schmidt CH, Ladda RL. (1985) Encouraging patients to undergo prenatal genetic counselling before the day of amniocentesis. J Reprod Med 30: 933-935.

Loukopoulos D. (1991) Screening for thalassaemia and HbS in the East Mediterranean. Am J Hum Genet 49(Supp): 79.

McDowell I, Newell C. (1987) Measuring Health - A Guide to Rating Scales and Questionnaires. Oxford University Press.

McFarlane of Llandaff (1991) The research process in nursing. The Future. In: Cormack D. (Ed) The Research Process in Nursing. Blackwell Scientific Publications. Second edition.

McGrath A, Reid N, Boore J. (1989) Occupational stress in nursing. Int J Nurs Studies 26(4): 343-358.

MacGregor E. (1990) Dilemmas in genetic screening. Nursing 4(41): 13-15.

McIntosh J (1989) Models of childbirth and social class: a study of 80 working class primigravidae. In: Robinson S, Thomson AM (eds) Midwives, research and childbirth. Chapman and Hall.

McIntyre S. (1987) Women's experiences and attitudes to screening. In: King's Fund Forum Screening for fetal and genetic abnormality Programme and abstracts.

MacKenzie IZ. (1992) Pregnancy termination. In: Brock DJH, Rodeck CH, Ferguson-Smith MA. (Eds) Prenatal Diagnosis and Screening. Churchill Livingstone, Edinburgh. page 675-685.

McKusick VA. (1990) Mendelian Inheritance in Man: Catalogs of autosomal dominant, autosomal recessive, and X-linked phenotypes, 9th ed (Baltimore MD: The Johns Hopkins University Press).

McMahon B. (1992) Positive use of a traumatic reawakening: Therapy with incest survivors. Professional Nurse 8(1):21-25.

Magni G, Salmi A, Paterlini A, Merlo A. (1982) Psychological distress in duodenal ulcer and acute gastroduodenitis. A controlled study. Digestive Diseases and Sciences 279(12):1081-1084.

Manchester University (1993) Department of Medical Genetics, St Mary's Hospital, Whitworth Place, Manchester M13 0JH, UK

Mander R. (1991a) Midwifery support of the mother relinquishing her baby for adoption: midwives' perceptions. Midw Chron & Nurs Notes October :275-284.

Mander R. (1991b) Midwifery care of the grieving mother: how the decisions are made. Midwifery 7:133-142.

Mant D, Fowler G. (1990) Mass screening:theory and ethics. BMJ 300:916-918.

Mari JDJ, Williams P. (1985) A comparison of the validity of two psychiatric screening questionnaires (GHQ-12 and SRQ-20) in Brazil using relative operating characteristics (ROC) analysis. Psychol Med 15: 651-659.

Marteau TM, Johnston M, Plenicar M, Shaw RW, Slack J. (1988a) Development of a self-administered questionnaire to measure women's knowledge of prenatal screening and diagnostic tests. J Psychosom Research 12: 403-408.

Marteau TM, Kidd J, Cook R, Johnston M, Michie S, Shaw R, Slack J. (1988b) Screening for Down's syndrome. (letter) BMJ 297: 1469.

Marteau TM. (1989a) Psychological costs of screening. BMJ 299:527.

Marteau TM, Johnston M, Shaw RW, Michie SM, Kidd J, New M. (1989b). The impact of prenatal screening and diagnostic testing upon the cognitions, emotions and behaviour of pregnant women. J Psychosom Res 33: 7-16.

Marteau TM, Kidd J, Cook R, Michie S, Johnston M, Slack J, Shaw RW. (1991a) Perceived risk not actual risk predicts uptake of amniocentesis. Br J Obst Gynaecol 98: 282-286.

Marteau TM. (1991b) Psychological implications of prenatal diagnosis. In: Drife JO, Donnai D. (Eds) Antenatal diagnosis of fetal abnormalities. Springer-Verlag, London.

Marteau TM, Slack J. (1992a) Psychological implications of prenatal diagnosis for patients and health professionals. In: Brock DJH, Rodeck CH, Ferguson-Smith MA. (Eds) Prenatal Diagnosis and Screening. Churchill Livingstone, Edinburgh.

Marteau TM, Van Duijn M, Ellis I. (1992b) Effects of genetic screening on perceptions of health: a pilot study. J Med Genet 29: 24-26

Marteau TM, Slack J, Kidd J, Shaw R. (1992c) Presenting a routine screening test in antenatal care: practice observed. Public Health 106:131-141.

Maskey S. (1991) Teenage pregnancy: doubts, uncertainties and psychiatric disturbance. J Roy Soc Med 84(12): 723-725.

Mennie ME, Holloway SM, Brock DJH. (1990) Attitudes of general practitioners to presymptomatic testing for Huntington's disease. J Med Genet 27: 224-227.

Mennie ME, Liston WA, Brock DJH. (1992a) Prenatal cystic fibrosis carrier testing: designing an information leaflet to meet the specific needs of the target population. J Med Genet 29: 308-312.

Mennie ME, Compton ME, Gilfillan A, Liston WA, Brock DJH (1992) Personal perception of CF carrier risk among 1,000 screened pregnant women. Unpublished data.

Mennie ME, Compton ME, Liston WA, Brock DJH. (1994) Gene therapy for cystic fibrosis: will it affect the uptake of screening. Prenatal Diagnosis (in press).

Menzies IEP. (1960) Nurses under stress. Int Nurs Review 7(6):9-16.

Merlin R. (1992) Understanding bulimia and its implications in pregnancy. JOGN 21(3): 199-205.

Merz B. (1987) Matchmaking scheme solves Tay-Sachs problem. J Am Med Assoc 258:2636-2639

Meyers D (1990) The family and life and death decisions. In Sutherland E, McCall Smith A. (Eds). Family Rights Family Law and Medical Advance. Edinburgh University Press

Michelson J, Gee S. (1984) In: Coming Late to Motherhood. Thorson Publishers Ltd.

Miller AE, Sliepcevich EM, Vitello EM. (1981) Health-related articles in the six leading women's magazines: content, coverage and readership profile. Health Values: Achieving High Level Wellness 5: 157-164

Modell B, Petrou M. (1988) Review of control programs and future trends in the United Kingdom. Birth Defects: Original Article Series 23(5b): 433-442. March of Dimes Birth Defects Foundation.

Modell B. (1992) Screening as public policy. In: Brock DJH, Rodeck CA, Ferguson-Smith MA. Prenatal diagnosis and screening.Churchill Livingstone, Edinburgh.

Modell B. (1990) Cystic fibrosis screening and community genetics. J Med Genet 27: 475-479.

Muylder de X. (1989) Psychological factors and preterm labour. J Reprod Infant Psychol 7: 55-57.

Mouzouros M, Camba L, Anionwu E. (1980) Thalassaemia as a model of recessive gentic disease in the community. Lancet ii: 574-578.

Newton R, Webster P, Binu P, Maskrey N, Phillips A. (1979) Psychosocial stress in pregnancy and its relation to the onset of premature labour. BMJ 2: 411-413.

Nott PN, Cutts S. (1982) Validation of the 30-item General Health Questionnaire in post partum women. Psychol Med 12: 409-413.

Nuffield Council on Bioethics (1993) Genetic screening and ethical issues.

Oakley A, McPherson A, Roberts H. (1984) Miscarriage. Penguin, London.

Oakley A, Rajan L, Grant A. (1990) Social support and pregnancy outcome. Br J Obstet Gynaecol 97:155-162.

Office of Health Economics (1986) Cystic fibrosis. London:OHE Cited in: Royal College of Physicians'Working Party. (1989) Report on prenatal diagnosis and genetic screening: community and service implications. Royal College of Physicians, London:RCP.

Office of Population Censuses and Surveys (1980) Classification of Occupations, HMSO London.

Oglethorpe RJL. (1989) Parenting after perinatal bereavement - a review of the literature. J Reprod Infant Psychol 7: 227-244.

Page H. (1988) The increasing demand for infertility treatment. Health Trends 4 (20): 115-118.

Parkes CM. (1975) Bereavement: Studies of grief in adult life. Penguin, Middlesex.

Patterson JM, Budd J, Goetz D, Warwick WJ. (1993) Family correlates of a 10-year pulmonary health trend in cystic fibrosis. Pediatrics 91(2): 383-389.

Paykel ES, Emms EM, Fletcher L, Rassaby E. (1980) Life events and social support in puerperal depression. Br J Psychiat 136:339-346.

Pearn JH. (1973) Patients' subjective interpretation of risks offered in genetic counselling J Med Genet 10:129-134.

Pembrey M (1987) Genetic and congenital disorders. In Screening for fetal and genetic abnormality. Kings Fund Forum.

Penkeith ARL, Wise A, Mearns M. (1987) CF in adolescents and adults. Thorax 42:526-532.

Phinn N (1990) Can economics be applied to prenatal screening? Discussion Paper 74.Centre for Health Economics, York. cited in: Clarke A (1993) Ethical issues raised by the 'new genetics'. In: Young ID (ed) The new Genetics, Ballierere's Clinical Paediatrics, Balliere Tindall, London.

Phipps S, Zinn AB. (1986) Psychological response to amniocentesis: I. Mood state and adaptation to pregnancy. Am J Med Genet 25: 131-142.

Pierce BA. (1990) Family Genetic Sourcebook. John Wiley & Sons, Inc. New York.

Polit DF, Hungler BP. (1989) Essentials of nursing research: methods, appraisal and utilization 2nd edition JB Lippincott Company, Philadelphia.

Pratt D. (1990) The partner's role in pregnancy. Nursing (UK) 4(9): 23-24.

Price B. (1990) Body Image: Nursing Concepts and Care. Prentice Hall International (UK) Ltd.

Pullen I. (1990) Patients, families and genetic information. In: Sutherland E, McCall McCall Smith A. (Eds) Family Rights Family Law and Medical Advance. Edinburgh University Press.

Quine L, Rutter DR, Gowen S. (1993). Women's satisfaction with the quality of the birth experience: a prospective study of social and psychological predictors. J Reprod Infant Psychol 11(2):107-113.

Rajan L, Oakley A. (1993) No pills for heartache: the importance of social support for women who suffer pregnancy loss. J Reprod Infant Psychol 11: 75-87.

Raphael-Leff J. (1991) Psychological processes of childbearing. Chapman Hall, London.

Rees C. (1990) The questionnaire in research. Nursing Standard 4(42): 34-35.

Reynolds BD, Puck M, Robinson A. (1974) Genetic counselling: an appraisal. Clin Genet 5: 177-187.

Richards M. (1987) Some reflections on fetal diagnosis. In: King's Fund Forum Screening for fetal and genetic abnormality Programme and Abstracts.

Richard MPM. (1989) Social and ethical problems of fetal diagnosis and screening. J Reprod Infant Psychol 7: 171-185.

Ridgeway V, Mathews A. (1982) psychological preparation for surgery:a comparison of methods. Br J Clin Psychol 21: 271-280.

Rigot JM, Lafitte JJ, Dumar V. (1991) Cystic fibrosis and congenital absence of the vas deferens. N Eng J Med 325: 64

Riordan JR, Rommens JM, Kerem B, Alon N, Rozmahel R, Grzelczak Z, Zielenski J, Lok S, Plavsic N, Chou J, Drumm ML, Iannuzzi MC, Collins FS, Tsui L. (1989) Identification of the cystic fibrosis gene: cloning and characterisation of complementary DNA. Science 245: 1066-1073.

Roberts L. (1990) One worked: the other didn't. (genetic screening programs for Tay-Sachs and sickle cell anaemia) Science 247:18.

Robinson J, Tennes K, Robinson A. (1975) Amniocentesis:its impact on mothers and infants. A 1-year follow-up study. Clin Genet 8: 97-106.

Robinson JO, Hibbard BM, Laurence KM. (1984) Anxiety during a crisis: emotional effects of of screening for neural tube defects. J Pychosom Res 28: 163-169.

Robinson S, Golden J, Bradley S. (1983) A study of the role and responsibilities of the midwife. Nursing Education Research Unit, Department of Nursing Studies, Chelsea College, University of London.

Rogers J, Matsumura M. (1991) Mother to be guide to pregnancy and birth for women with disabilities. Demos Publications, New York.

Rommens JM, Ianuzzi MC, Kerem B, Drumm Ml, Melmer G, Dean M, Rozmahel R, Cole JL, Kennedy D, Hidaka N, Zsiga M, Buckwald M, Riordan JR, Tsui LC, Collins FS. (1989) Identification of the cystic fibrosis gene: Chromosome walking and jumping. Science 245: 1059-1065.

Rosenstock IM, Child B, Simopoulous AP. (1975) Knowledge attitude and behaviour. In: Genetic screening: programs, principles and research. Washington DC: National Accademy of Sciences.

Rotheram J. (1989) Care of the disabled woman during pregnancy. Nurs Stand 4(10): 36-39.

Rothman Katz B (1988) The Tentative Pregnancy. Penguin, New York.

Rowley PT. (1984) Genetic screening: Marvel or menace? Science 225: 138-144.

Rowley PT, Loader S, Sutera CJ, Walden M, Kozyra A. (1991) A prospective regional trial of Prenatal screening for hemoglobinopathies. I. . A m J Hum Genet 48: 439-446.

Royal College of Midwives, London (1981). Practical guidance for midwives facing ethical or moral dilemmas.

Royal College of Physicians'Working Party. (1989) Report on prenatal diagnosis and genetic screening: community and service implications. Royal College of Physicians,London:RCP.

Report of Royal College of Physicians, London (1990) Cystic fibrosis in adults.

Royal College of Physicians, London (1993) National confidential enquiry into counselling for genetic disorders. Cited in: Midwives Chronicle (1993) 106 (1264): 164.

Saiki RK, Gelfand DH, Stoffel S. (1988) Primer directed enzymatic amplification of DNA with a thermostable DNA polymerase. Science 239: 487-491.

Sandelowski M, Harris BG, Holditch-Davis D. (1991) Amniocentesis in the context of infertility. Health Care for Women International 12: 167-168.

Sanden ML. (1985). Midwives knowledge of the alpha-fetoprotein test. J Psychosom Obstet Gynaecol 4:23-30.

Sanden ML, Bjurulf P. (1988) Pregnant women's attitudes for accepting or declining a serum-alpha-fetoprotein test. Scand J Soc Med 16: 265-271.

Schuman AN, Marteau TM. (1993) Obstetricians' and midwives' contrasting perceptions of pregnancy. J Reprod Infant Psychol 11(2): 115-118.

Scottish Home and Health Department (1983) Shared Care in Obstetrics: report by working group of national medical consultative committee.

Scottish Home and Health Department Scottish Health Service Advisory Council (1991) Report of the Study Group on the Management of Gynaecological Services in Scotland. HMSO, Edinburgh.

Scottish Home and Health Department (1993) Provision of Maternity Services in Scotland. A Policy Review. HMSO, Edinburgh.

Scriver CR, Clow CL. (1990) Carrier screening for Tay-Sachs disease. Lancet 336: 191

Seyle H. (1974) Stress Without Distress. Hodder and Stoughton, London.

Shapiro DA, Shapiro LR. (1989) Pitfalls in Tay-Sachs carrier detection: Physician referral patterns and patient ignorance. N.Y. State J Med 89: 317-319.

Sharp DJ. (1988) Validation of the 30-item General Health Questionnare. Psychol Med 18: 503-507.

Shepherd SL, Melbourne FH, Harwood IR, Granger LE, Hofstetter CR, Molgaars C, Kaplan RM. (1990) A comparative study of the psycho social assets of adults with cystic fibrosis and their healthy peers. Chest 97: 1310-1316

Shershevsky PM, Yarrow LM. (Eds) (1973) Psychological aspects of a first pregnancy and early postnatal adaptation. Raven Press, New York.

Shiloh S, Sagi M (1989) Effect of framing on the perception of genetic recurrence risks. Am J Med Genet 33:130-135.

Shiloh S, Avdor O, Goodman RM. (1990) Satisfaction with genetic counselling: Dimensions and measurements. Am J Med Genet 37:522-529.

Shrimpton AE, McIntosh I, Brock DJH. (1991) The incidence of different cystic fibrosis mutations in the Scottish population: Effects on prenatal diagnosis and genetic counselling. J Med Genet 28: 317-321.

Simpson JL. (1991) Screening for fetal and genetic abnormalities. In: Walters WAW. (Ed) Clinical obstetrics and gynaecology international practice and research. Human reproduction: current and future ethical issues. Bailliere Tindall. London.

Sjogren B, Uddenberg N. (1990) Prenatal diagnosis for psychological reasons: comparison with other indications, advanced maternal age and known genetic risk. Prenat Diagn 10: 111-120.

Sjogren B. (1992) Future use and development of prenatal diagnosis. Consumers' attitudes. Prenat Diag 12: 1-8.

Smith M, Treadwell M, O'Grady L. (1983) Psychosocial functioning, life change and clinical status in adolescents with cystic fibrosis. J Adolesc Health Care 4: 1310-1316.

Spielberger CD, Gorsuch RL, Luchene R, Vagg PR, Jacobs GA. (1983) Manual for the State-Trait Anxiety Inventory. Consulting Psychologists Press, Palo Alto. Cited in: Thompson C. (Ed) (1989). The instruments of psychiatric research. John Wiley & Sons Ltd.

Stafford EM, Jackson PR, Banks MH. (1980) Employment, work involvement and mental health in less qualified young people. J Occup Psychol 53: 291-304.

Stewart A. (1992) Creating happy families. The effects of adoption on those involved. Professional Nurse 7(8): 497-505.

Stirk F. (1991) Sickle cell in pregnancy. Nurs Times 87(41):36-38.

Stoate HG. (1989) Can health screening damage your health? J R Coll Gen Pract 39: 193-195.

Statham H, Green J. (1993) Serum screening for Down's syndrome: some women's experiences. BMJ 307: 174-176.

Stringer M, Librizzi R, Weiner S. (1991) Establishing a prenatal genetic diagnosis: the nurses role. Mat Child Nurs 16: 152-156.

Sullivan LW. (1987) Risks of sickle-cell trait: caution and common sense. N Eng J Med 317: 830-831.

Super M, Schwarz MJ, Malone G. (1992) Screening for cystic fibrosis carriers Lancet 340:490-491.

Sutherland VJ, Cooper CL. (1990) Understanding stress. A psychological perspective for health professionals. Chapman and Hall, London.

Sutherland E. (1990) Regulating pregnancy: should we and can we? In: Sutherland E, McCall Smith A. (Eds) Family Rights Family Law and Medical Advance. Edinburgh University Press.

Sutton A. (1990) Prenatal diagnosis: confronting the ethical issues. The Linacre Centre, London.

Sweet BR. (1988) Mayes' Midwifery. A textbook for midwives. 11th Edition. Bailliere Tindall, London

Tabor A, Norgaard-Pedersem B, Jacobson JC. (1984) Low maternal serum AFP and Down Syndrome. Lancet 2(8395): 161-162.

Tabor A, Jonsson MH. (1987) Psychological impact of amniocentesis in low risk women. Prenat Diag 7: 443-449.

Targum SD. (1981) Psychotherapeutic considerations in genetic counselling. Am J Med Genet 8: 281-289.

Taylor C. (1993) Nutrition and the child with cystic fibrosis. Paed Nurs 5(4): 26-28.

Tesch R. (1991) Software for qualitative researchers: analysis needs and program capabilities. In: Fielding NG, Lee RM. Using Computers in Qualitative Research. Sage Publications, London.

Tierney AJ, Taylor J. (1991) Research in practice: an experiment in researcher-practitioner collaboration. J Adv Nurs 16:506-510.

Tinsley BJ, Trupin SR, Owens L, Boyum LA. (1993) The significance of women's pregnancy-related locus of control beliefs for adherence to recommended prenatal health regimes and pregnancy

Titchen A, Binnie A. (1993) Research partnerships: collaborative action research in nursing. J Adv Nurs 18: 858-865.

The National Health Service for Scotland 1991-92 Framework for Action Impact on NHS staff. HMSO.

Thompson C. (1989) Self-Rating Scales for Anxiety. In:Thompson C. (ed) The Instruments of Psychiatric Research. John Wiley and Sons Ltd.

Thornley CN, Walton VA, Romans-Clarkson SE, Herbison GP, Mullen PE. (1991). Screening for psychiatric morbidity in men and women. N.Z. Med J. 104(925): 505-507.

Thorp JM Jr, Bowes WA Jr. (1989) Prenatal diagnosis for couples who would not consider abortion. (Letter) Obstet Gynecol 74:828-829

Trethowan WH, Conlon MF. (1965) The couvade syndrome. Br J Psychiat 111: 57-66.

Trethowan WH. (1968) The couvade syndrome some further observations. J Psychosom Research 12: 19-16.

Trezise AE, Buchwald M. (1991) In vivo cell specific expression of the cystic fibrosis transmembrane conductance regulator. Nature 353: 434-437.

Tsoi MM, Hunter M. (1987a) Ultrasound scanning in pregnancy: Consumer reactions. J Reprod Infant Psychol 5: 543-548.

Tsoi MM, Hunter M, Pearce M, Chudleigh P, Campbell S. (1987b) Ultrasound scanning in women with raised serum alpha-fetoprotein:short term psychological effects. J Psychosom Research 31: 35-39.

Tsui-der Chou G. (1986) dBase III Plus Handbook 2nd edit QUE Corporation, Indianapolis.

Tunis SL, Golbus MS. (1991) Assessing mood states in pregnancy:survey of the literature. Obstet Gynecol Survey 46:340-346.

Tymstra TJ. (1989) The imperative character of medical technology and the meaning of anticipated decision regret. Int J Tech Asess in Health Care 5: 209-213.

Tymstra TJ, Bajema C, Beekhuis JR, Mantingh A. (1991) Women's opinions on the offer and use of prenatal diagnosis. Prenat Diagn 11: 893-898.

United States Congress Office of Technology Assessment (1992a) Cystic fibrosis and DNA tests: implications of carrier screening.

United States Congress, Office of Technology Assessment (1992b) Cystic Fibrosis and genetic screening: policies, practices and attitudes of gentic counsellors - results of a survey. Government Printing Office Washington DC.

Van Cleve L. (1993) Alpha-fetoprotein testing:opinions of parents of children with spina bifida. Mat-Child Nurs J 21(1):20-26.

Van den Berghe H. (1987) Impact of genetics on society. In: Evers-Kiebooms G,

Cassiman JJ, Van den Berghe H, d'Ydewalle G. (Eds) Genetic risk, risk perception and decision making. Birth Defects Original Article Series 23: 1-5.

Verjaal M, Leschot NJ, Treffers PE. (1982) Women's experiences with second trimester prenatal diagnosis. Prenat Diag 2:195-209.

Vinson PC, Finley SC, Davis RO, Huggins WH, Rigdon DT, Finley WH. (1980) Amniocentesis for prenatal genetic studies: a reassuring experience. Am J Hum Genet: 32: 135A.

Vuori H. (1989) Research needs in quality assurance. Quality assurance in health care 1;2/3:147-159

Wald N, Cuckle H. (1988) AFP and age screening for Down syndrome Am J Med Genet 31:197-209.

Walters S, Britton J, Hodson ME. (1993) Demographic and social characteristics of adults with cystic fibrosis in the United Kingdom. BMJ 306: 549-552.

Watson E, Evans S. (1986) An example of cross-cultural measurement of psychological symptoms in post-partum mothers. Soc Sci Med 23: 869-874.

Watson JP, Elliott SA, Rugg AJ, Brough ID. (1984) Psychiatric disorder in pregnancy and the first postnatal year. Br J Psychiat 144: 453-462.

Watson EK, Mayall E, Chapple J, Williamson R. (1991) Screening for carriers of cystic fibrosis through primary health care services. BMJ 303:504-507.

Watson EK, Mayall E, Chapple J, Williamson R. (1992) Psychological and social consequences of a community carrier screening programme for cystic fibrosis. Lancet 340:217-220.

Weatherall DJ. (1991a) The New Genetics and Clinical Practice.(3rd edition) Oxford University Press.

Weatherall DJ. (1991b) The evolution of thalassaemia screening. Am J Hum Genet 49(Supp):79.

Weekend Financial Times (1992) Changing face of the UK. Saturday 1st February.

Wertz DC, Sorenson JR, Heeren TC. (1984) Genetic counseling and reproductive uncertainty. Am J Med Genet 18: 79-88.

Wertz DC, Fletcher JC. (1990) International perspectives on voluntary versus mandatory screening and third party accesss to tests results. In: Knoppers BM; Laberge CM. (Eds) Genetic screening: from newborns to DNA typing. Excerpta Medica, New York.

Whelton J. (1989) The midwife and foetal medicine. Nursing (Dec) 8-

White-Van Mourik MCA, Connor JM, Fergerguson-Smith MA. (1992) The psychological sequelae of a second-trimester termination of pregnancy for fetal abnormality. Prenat Diagn 12: 189-204.

Whitten CF (1993) Sickle-cell programming - an imperilled promise New Eng J Med 288:318-319

WHO Expert Committee on Human Genetics (1969) Genetic counselling. Wld Hlth Org. Techn. Rep. Ser. 416:1-23

Whyte DA. (1992) A family nursing approach to the care of a child with a chronic illness. J Adv Nurs 17:317-327.

Wilday J. (1989) Ethics in Midwifery. Midwives' Chronicle 176.

Wilfond BS, Fost N. (1990) The cystic fibrosis gene: medical and social implications for heterozygote detection. JAMA 263:2777-2783.

Wilkie T. (1992) This mouthwash could mean a lot to your children. The Independent, Science Section, 20th January page 14.

Wilkinson G. (1992) In: Anxiety: recognition and treatment in general practice. Radcliffe Medical Press.

Williamson R, Allison MED, Bentley TJ, Lim SMC, Watson E, Chapple J, Adam S, Boulton M. (1989) Community attitudes to cystic fibrosis carrier testing in England: a pilot study. Prenat Diagn 9: 727-734.

Wilson-Barnett J. (1988) Patient teaching or patient counselling? J Adv Nurs 13: 215-222.

Wolff JRF, Nielson PE, Schiller P. (1970) The emotional reaction to stillbirth. Am J Obstet Gynecol 108:73-76.

Wolfkind S, Zajicek E. (1981) Pregnancy: A Psychological and Social Study. Academic Press, London.

Wrightson N, Thompson P, Blake A. (1993) Breathing new life. Nurs Times 89(23):38-41.

Younger JB. (1991) A model of parenting stress. Research in Nursing & Health 14:197-204.

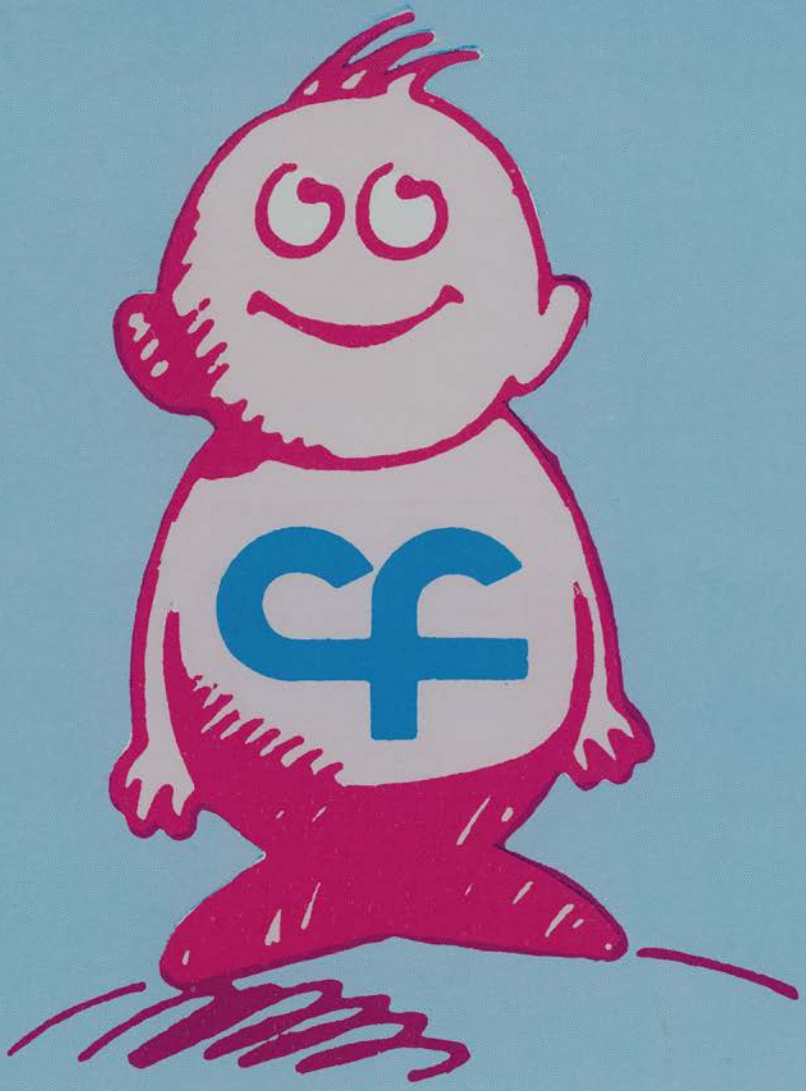
Zalk SR. (1980) Psychosexual conflicts in expectant fathers. In: Blum BL. (Ed) Psychological aspects of pregnancy, birthing and bonding. Human Sciences Press, New York.

Zeesman S, Clow CL, Cartier L, Scriver CR. (1984) A private view of heterozygoygosity: eight year follw-up study on carriers of the Tay-Sachs gene detected by high school screening in Montreal. Am J Med Genet 18:769-778.

Zhang JZ, Cai SP, Khan YW. (1991) Thalassaemia screening and prenatal diagnosis in South China. Am J Hum Genet 49(Supp):80.

APPENDIX

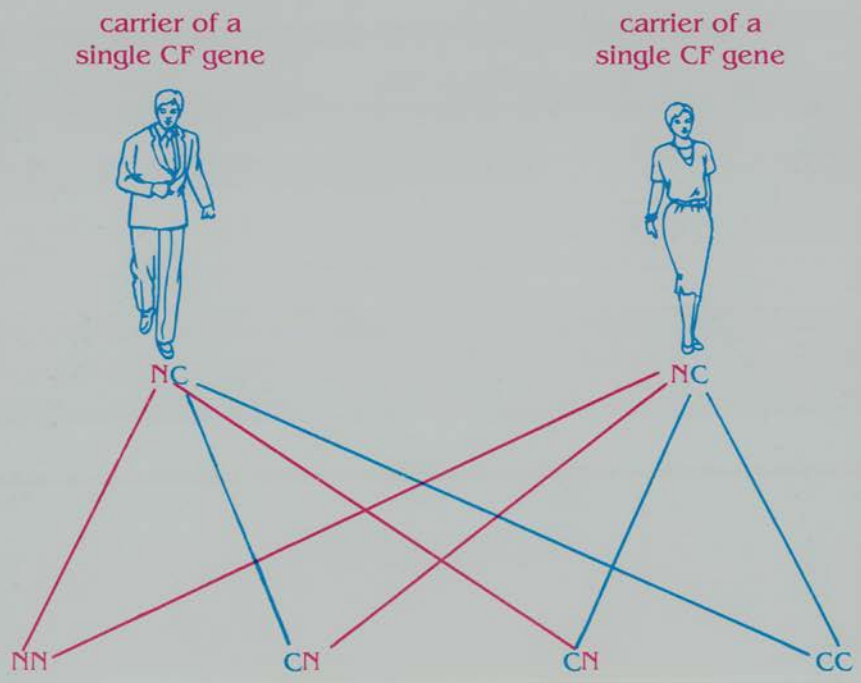
CYSTIC FIBROSIS CARRIER TESTING



PRENATAL SCREENING TRIAL

One in every 25 men and women carry a single cystic fibrosis gene (CF gene). A single CF gene is completely harmless. But if a woman who carries a single CF gene has a child by a man who also carries a single CF gene, there is a 1 in 4 chance that their child will have the disease cystic fibrosis (CF).

AT EACH PREGNANCY, THIS COUPLE HAS A 1 IN 4 CHANCE OF HAVING A CHILD WITH CYSTIC FIBROSIS



N = normal gene
C = CF gene

THE DISEASE – CYSTIC FIBROSIS

CF is serious. The average life expectancy is 25 years. CF causes thick mucus to gather in the lungs and gut. Children with CF need chest physiotherapy several times a day and antibiotics to fight chest infections. If they live long enough they will probably need a heart and lung transplant.

Children with CF require a balanced diet, with 50% more food than the average child their age and tablets given daily to help digestion.

MOST CHILDREN WITH CF ARE BORN TO PARENTS WHO HAVE NO FAMILY HISTORY OF THE DISEASE

In 1 in 600 couples, both partners carry a single CF gene. Most of these couples have no family history of the disease and are unaware they have a 1 in 4 chance of having a child with CF. Even if you already have healthy children, you may still be a CF carrier.

WE THINK THAT A WOMAN WHO IS HAVING A BABY WILL WANT TO KNOW THE ANSWERS TO THREE IMPORTANT QUESTIONS

1. AM I A CF CARRIER?

2. IS MY PARTNER A CF CARRIER?

3. COULD MY BABY HAVE CF?

THE CYSTIC FIBROSIS CARRIER SCREENING TEST

A mouthwash sample is used to test if you are a CF carrier. You will be contacted in 1 week, only if the test shows you are a carrier.

Testing for the CF gene is successful in 85% of cases. This means we cannot guarantee that you will not have a child with CF. However, if the test is negative it will greatly reduce the risk.

If you wish your negative test result, bring a S.A.E. to the clinic.

TESTING YOUR PARTNER

If you are found to carry a single CF gene we would be able to test your partner.

He also has a 1 in 25 chance of being a CF carrier.

If your partner's test is negative then your risk of having a baby with CF is very low.

A special leaflet and genetic counselling are available for all couples where one partner only has a positive carrier test.

If however, you don't know who the father of your baby is, we advise against taking the CF carrier test.

PRENATAL CARRIER TESTING FOR CYSTIC FIBROSIS

I .....

WISH TO PARTICIPATE IN THE CYSTIC FIBROSIS CARRIER TESTING SCREENING TRIAL.

I UNDERSTAND THAT THE CARRIER TEST FOR THE CYSTIC FIBROSIS GENE DOES NOT GUARANTEE THAT I WILL NOT HAVE A CHILD WITH CYSTIC FIBROSIS.

I UNDERSTAND THAT I CAN WITHDRAW FROM THE SCREENING TRIAL AT ANY TIME.

SIGNED .....

DATE .....

COUPLES WHO ARE BOTH CARRIERS OF A SINGLE CYSTIC FIBROSIS GENE

Genetic counselling is available to couples where both partners are found to be CF carriers.

The various options open to you will be fully explained. You will have time to think through these options and discuss them at length with the counsellor.

One option is prenatal diagnosis. It is possible to diagnose CF in the fetus. If the prenatal diagnostic test shows that the unborn child is going to have CF, you may wish to have a termination.

Prenatal diagnosis is not recommended for those couples who find termination unacceptable.

FURTHER INFORMATION AND ADVICE ABOUT PRENATAL CARRIER TESTING FOR CYSTIC FIBROSIS

There will be time at the antenatal booking clinic to ask questions, or discuss concerns you may have about prenatal carrier testing for cystic fibrosis. Counselling is available at any stage of the screening trial. A nurse counsellor is available at the antenatal booking clinic.

We think it is important that we find out a number of things about screening for CF during pregnancy. Firstly, are we giving you the information you need in our leaflet? Are we causing you any upset by screening you during pregnancy? Do you and your partner find this an acceptable time to be screened? The enclosed questionnaire is designed to answer these questions and help us give you the service you need. Please complete it before you come along to the clinic and give it to the midwife who is caring for you at the clinic.

PRENATAL CARRIER TESTING FOR CYSTIC FIBROSIS IS VOLUNTARY

Prenatal carrier testing for CF is entirely voluntary. If you would like to be tested please complete the enclosed consent form and bring it with you to the antenatal booking clinic. You can, of course, withdraw at any time from the screening trial.

IMPORTANT POINTS TO REMEMBER ABOUT PRENATAL CARRIER TESTING FOR CYSTIC FIBROSIS

1. Carrying a single CF gene is completely harmless.
2. Only if both partners carry a single CF gene will there be a 1 in 4 chance that their child will have the disease CF.
3. Most children are born to couples who have no family history of the disease.
4. If you don't know who the father of your baby is, we advise against taking the CF carrier test.
5. Testing for the CF gene is still only successful in 85% of cases.
6. Genetic counselling is available for all couples where both partners are found to carry a single CF gene.
7. The prenatal cystic fibrosis carrier screening trial is entirely voluntary.
8. Remember there are trained staff to give you information and advice – please do ask.

Printed and Designed by the University of Edinburgh

CYSTIC FIBROSIS CARRIER TESTING

PRENATAL SCREENING TRIAL

The cystic fibrosis carrier screening test is new. This is why we are running a prenatal screening trial. You can tell us if pregnancy is a good or a bad time to be offering you this test.

This questionnaire is designed to answer two important questions. Firstly, are we giving you and your partner the information you need to help you make up your mind whether or not to have the CF screening test? Secondly, are we causing you any upset by testing you when you are pregnant?

Please fill in the questionnaire at home and bring it with you to the booking clinic. The midwife looking after you at the clinic will collect it from you. If you have decided NOT to have the CF carrier test, or are UNDECIDED we would still like you to fill in the questionnaire.

THANK YOU FOR YOUR HELP.

Please tick the appropriate box or boxes.

1. Had you heard of cystic fibrosis, before you read the CF leaflet?

   a) yes ☐ b) no ☐ c) don't know ☐

2. If you had heard of cystic fibrosis, where did you hear about it?
   (Tick as many boxes as apply)

   a) television ☐ b) newspaper ☐ c) radio ☐

   d) women's journal ☐ e) charity appeal ☐ f) work ☐

   g) other ☐ h) can't remember ☐

3. Is the leaflet describing the CF carrier test?

   a) difficult to understand ☐ b) easy to understand ☐

4. Has your partner read the leaflet?

   a) yes ☐ b) no ☐ c) don't know ☐

5. If he read it did he find it?

   a) difficult to understand ☐ b) easy to understand ☐ c) don't know ☐

6. Do you know what your chance of carrying a single CF gene is?

   a) 1 in 4 ☐ b) 1 in 25 ☐ c) 1 in 100 ☐

   d) 1 in 200 ☐ e) Don't know ☐

7. Have you discussed the CF carrier test with anyone?

   a) partner ☐ b) relative ☐

   c) friend ☐ d) GP ☐

   e) health visitor ☐ f) other (state who) ☐

   g) no-one ☐

8. Have you decided?

a) to have the test ☐

b) not to have the test ☐

c) not decided ☐

9. In a few words can you say why you have made this decision.
(please write your reasons in the space below)

10. Does the thought of the CF test make you feel?

a) anxious ☐

b) reassured ☐

c) don't know ☐

11. Do you think the leaflet should give you more information?

a) yes ☐ b) no ☐ c) don't know ☐

12. If yes, what would you like to know?

a) I'd like to know more about the disease CF ☐

b) I'd like to know more about the treatment for CF ☐

c) I'd like to know more about how CF is passed on to children ☐

d) I'd like to know more about the CF carrier screening test ☐

e) I'd like to know more about diagnosing CF in the unborn child ☐

f) I'd like to know other information (please state) ☐

PLEASE TURN OVER

GENERAL HEALTH QUESTIONNAIRE

PLEASE READ THIS CAREFULLY - We should like to know how you have been feeling generally over the past few weeks. Please answer all the questions by circling the answer which you think most nearly applies to you.

REMEMBER - we want to know about present or recent complaints, not those you have had in the past.

It is important that you try to answer ALL the questions.

HAVE YOU RECENTLY:

1 - been able to concentrate on whatever you're doing?	better than usual	same as usual	less than usual	much less than usual
2 - lost much sleep over worry?	not at all	no more than usual	rather more than usual	much more than usual
3 - felt that you are playing a useful part in things?	more so than usual	same as usual	less useful than usual	much less useful
4 - felt capable of making decisions about things?	more so than usual	same as usual	less so than usual	much less capable
5 - Felt constantly under strain?	not at all	no more than usual	rather more than usual	much more than usual
6 - felt you couldn't overcome your difficulties?	not at all	no more than usual	rather more than usual	much more than usual
7 - been able to enjoy your normal day-to-day activities?	more so than usual	same as usual	less so than usual	much less than usual
8 - been able to face up to your problems?	more so than usual	same as usual	less able than usual	much less able
9 - been feeling unhappy and depressed?	not at all	no more than usual	rather more than usual	much more than usual
10 - been losing confidence in yourself?	not at all	no more than usual	rather more than usual	much more than usual
11 - been thinking of yourself as a worthless person?	not at all	no more than usual	rather more than usual	much more than usual
12 - been feeling reasonably happy all things considered?	more so than usual	about same as usual	less so than usual	much less than usual

---

Thank you for taking the trouble to fill in this questionnaire

REMEMBER - to bring the questionnaire with you to the clinic

CYSTIC FIBROSIS CARRIER TESTING
PRENATAL SCREENING TRIAL

Describe how you have felt during the PAST WEEK
if you have not had the symptoms at all make a check mark (√) in the box on the left like :

	Not at all	A little slightly	A great deal, Quite a bit	Extremely could not have been worse
Headaches or head pains	√			

If you have had the symptoms describe how much it has bothered you or troubled you, for example, like this:

	Not at all	A little slightly	A great deal Quite a bit	Extremely could not have been worse
Headaches or head pains			√	

Please answer all questions. Do not think long before answering.

	Not at all	A little, slightly	A great deal, Quite a bit	Extreme Could no have bee worse
1 Feeling dizzy or faint				
2 Feeling tired or a lack of energy				
3 Nervous				
4 Feelings of pressure or tightness in head or body				
5 Scared or frightened				
6 Poor appetite				
7 Heart beating quickly or strongly without reason (throbbing or pounding)				
8 Feeling that there was no hope				
9 Restless or jumpy				
10 Poor memory				
11 Chest pains or breathing difficulties or feeling of not having enough air				
12 Feeling guilty				
13 Worrying				
14 Muscle pains, aches or rheumatism				
15 Feeling that people look down on you or think badly of you				
16 Trembling or shaking				
17 Difficulty in thinking clearly or difficulty in making up your mind				
18 Feeling unworthy or a failure				
19 Feeling tense or "wound up"				
20 Feeling inferior to other people				
21 Parts of body feel numb or tingling				
22 Irritable				
23 Thoughts which you cannot push out of your mind				
24 Lost interest in most things				
25 Unhappy or depressed				
26 Attacks of panic				
27 Parts of your body feel weak				
28 Cannot concentrate				
29 It takes a long time to fall asleep, or restless sleep or nightmares				
30 Awakening too early and not being able to fall asleep again				

CYSTIC FIBROSIS CARRIER TESTING
PRENATAL SCREENING TRIAL

FACTS AND FEELINGS QUESTIONNAIRE

FACTS QUESTIONNAIRE

Here are a number of statements, please tick the ones you think are true.

1 any couple can have a child with cystic fibrosis

true ☐

2 a couple can have a child with cystic fibrosis if

a) only one partner carries a single cystic fibrosis gene true ☐

b) both partners carry a single cystic fibrosis gene true ☐

3 if both partners carry a single cystic fibrosis gene their chance of having a child with the disease cystic fibrosis is

a) 1 in 2 true ☐ b) 1 in 4 true ☐

c) 1 in 20 true ☐

d) all their children will have cystic fibrosis true ☐

4 one in 25 people in Britain carry a single cystic fibrosis gene

true ☐

5 if you carry a single cystic fibrosis gene this means that

a) your health will be affected true ☐

b) you will develop the disease cystic fibrosis true ☐

c) it is only important if your partner also carries a single cystic fibrosis gene true ☐

6 even, if you have no family history of cystic fibrosis you can carry a single cystic fibrosis gene

true ☐

FEELINGS QUESTIONNAIRE

please tick which statement is CLOSEST to the way you feel

feel the information I was given about the cystic fibrosis
arrier test was

	not enough	not quite enough	about right	too much	much too much
before the antenatal booking clinic					
at the antenatal booking clinic					

feel I understood what the cystic fibrosis carrier test was all
bout before I was tested

yes ☐

no ☐

don't know ☐

am glad I had the cystic fibrosis carrier test

yes ☐

no ☐

don't know ☐

10 I feel I am in favour of testing for cystic fibrosis carriers

	yes	no	don't know
a) in schools			
b) in family planning clinics			
c) in GP health centres			

11 I feel that the cystic fibrosis carrier test should be routinely offered to pregnant women

yes ☐

no ☐

don't know ☐

12 if I turned out to carry a single cystic fibrosis gene, I feel I would tell

	yes	no	don't know
a) my partner			
b) my brothers and sisters			
c) my children			
d) my other relatives			
e) my friends			

thank you for filling in this questionnaire

PRENATAL DIAGNOSIS, VOL. 13, 807–814 (1993)

PRENATAL CYSTIC FIBROSIS CARRIER SCREENING: FACTORS IN A WOMAN'S DECISION TO DECLINE TESTING

M. E. MENNIE, A. GILFILLAN, M. E. COMPTON, W. A. LISTON* AND D. J. H. BROCK

*Human Genetics Unit, Department of Medicine, University of Edinburgh, Western General Hospital, Edinburgh EH4 2XU, U.K.; *Simpson Memorial Maternity Pavilion, Edinburgh, U.K.*

SUMMARY

Among 2207 women eligible to be screened for cystic fibrosis (CF) carrier status during pregnancy, 325 (15 per cent) declined to be tested. Of these, 260 (80 per cent) answered a questionnaire soliciting their reasons for not participating. The main factor was opposition to termination of pregnancy, with 43 per cent being against termination for any reason and another 11 per cent against termination of a CF fetus. Other reasons given were partner's disapproval or non-participation (10 per cent), perceived risk of a CF child being low (7 per cent), the error rate of the test (6 per cent), and the generation of unacceptable levels of anxiety (5 per cent). Eleven women (4 per cent) said that they did not wish to be tested during pregnancy, but only six of these would have accepted screening at another time.

KEY WORDS Cystic fibrosis Prenatal carrier screening Decision-making

INTRODUCTION

The main objective of cystic fibrosis (CF) carrier screening during pregnancy is to identify high-risk couples and to allow them to avoid the birth of an affected child through prenatal diagnosis. For the past 2 years, we have conducted a pilot trial of CF carrier screening in the major Edinburgh maternity hospital. Details of the information leaflet inviting participation and the protocol used have been published elsewhere (Mennie *et al.*, 1992a,b). During this period, about 15 per cent of women declined to enter the trial. This study examines the reasons given by women who have not wished to be screened during pregnancy.

SUBJECTS AND METHODS

Between May 1991 and January 1992, a total of 2541 women booking to have their babies at the Simpson Memorial Maternity Pavilion, Edinburgh were invited to enter the CF carrier screening trial. Details of the recruitment method are given elsewhere (Mennie *et al.*, 1992b). Of these, 334 (13 per cent) were not eligible for reasons given (Mennie *et al.*, 1992b). A total of 2207 (87 per cent) women met the criteria for screening, of whom 325 (15 per cent) declined and 1882 (85 per cent) were screened. Those who declined screening are the subjects of this study.

Addressee for correspondence: Moira E. Mennie, Human Genetics Unit, Department of Medicine, University of Edinburgh, Western General Hospital, Edinburgh EH4 2XU, U.K.

0197–3851/93/090807–08$09.00

Received 21 December 1992
Accepted 31 January 1993

Table 1. Back cover of the CF carrier test information leaflet summarizing the important points of screening

IMPORTANT POINTS TO REMEMBER ABOUT PRENATAL CARRIER TESTING FOR CYSTIC FIBROSIS
1. Carrying a CF gene is completely harmless
2. Only if both partners carry a single CF gene will there be a 1 in 4 chance that their child will have the disease CF
3. Most children are born to couples who have no family history of the disease
4. If you don't know who the father of your baby is, we advise against taking the CF carrier test
5. Testing for the CF gene is still only successful in 85 per cent of cases
6. Genetic counselling is available for all couples where both partners are found to carry a single CF gene
7. The prenatal cystic fibrosis carrier screening trial is entirely voluntary
8. Remember there are trained staff to give you information and advice—please do ask

Women were invited to enter the trial by means of an information leaflet sent with their antenatal booking appointment. The designing of the leaflet is described elsewhere (Mennie *et al.*, 1992a). The leaflet briefly acquainted women and their partners with the disease cystic fibrosis and the mode of inheritance. It outlined the purpose of prenatal CF carrier screening and explained the testing procedure. The leaflet emphasized that the test was entirely voluntary, was 85 per cent successful, and that a negative test result did not guarantee that a child would not have CF, but would greatly reduce the risk. It further emphasized that most couples who had a child with CF had no previous family history of the disease. The leaflet also stated that if a woman did not know who the father of her baby was, then it was inadvisable to take the test.

Prenatal diagnosis was discussed as an option for couples where both partners were CF carriers, and termination of pregnancy was cited as a further option if the prenatal diagnostic test showed that the baby had the disease CF. At this adjunct, it was stressed that prenatal diagnosis was not recommended for couples who found termination of pregnancy unacceptable. The back cover of the leaflet highlighted the important points for patients to note (see Table 1).

A self-administered prescreening questionnaire was included with the information leaflet. Women were asked to complete this at home and bring it with them to the clinic. The questionnaire asked if a woman had (a) decided to have the test, (b) decided not to have the test, or (c) not decided. It further asked if they would write in a few words why they had made this decision or remained undecided. Quotations were transcribed to a computer database and examined word by word to abstract meanings or themes, which were categorized and then coded (Field and Morse, 1990). The evaluation of the transcribed quotations and categories was independently carried out by two research colleagues.

Socio-demographic data including details of a woman's decision to participate in the alpha-fetoprotein (AFP) screening programme were obtained from the antenatal records.

RESULTS

At booking, 1812 (82 per cent) of the 2207 women eligible for screening had decided to be tested, of whom 1728 returned a questionnaire. Two hundred and seventy-nine (13 per cent) of the women declined the test, of whom 214 returned a questionnaire commenting on why they had made their decision. A further group of 116 (5 per cent) were undecided about the test at booking. Those who were undecided were counselled by a genetic nurse and commented on why they were uncertain. Of this group, 70 (60 per cent) were subsequently screened and 46 (40 per cent) declined. Questionnaires were therefore completed by 1798 women accepting and 260 women declining screening.

Demographic data showed that multiparous women (68 per cent) were more likely to decline CF carrier testing than primiparous women (60 per cent) (χ^2 $p<0{\cdot}05$; Table 2). Women who declined CF screening were also very much more likely to decline AFP screening (χ^2 $p<0{\cdot}001$). There were no other significant differences in socio-demographic data on the accepting and declining groups.

Two hundred and fourteen women commented on why they had declined the test and 46 who were initially uncertain and subsequently declined also commented on the reasons for their uncertainty. The categories derived from these comments are listed in Table 3 and examples of quotations are given in Table 4.

One hundred and forty (54 per cent) women declined to enter the trial because they did not wish to terminate the pregnancy. The majority of this category (111) were completely opposed to termination of pregnancy. However, 29 stated that they held this opinion specifically with regard to the disorder CF.

The CF carrier test is distinct from other prenatal screening tests with respect to involving the male partner. Indeed, prior indication that he would be willing to be tested if necessary was paramount. Twenty-six (10 per cent) of those who declined screening did so because their partner had indicated his reluctance to participate.

Perceiving one's risk of having an affected child as low clearly influenced women to decline the test. Nineteen (7 per cent) stated this to be their reason for refusing to be involved in the trial. Of these, 14 were multiparous women.

The inability of the CF carrier test to detect all carriers was a factor which influenced 15 (6 per cent) to refuse screening. This reason frequently overlapped with their concern that an inconclusive test result would generate anxiety throughout the remainder of the pregnancy.

Thirteen (15 per cent) of the women stated that the test would generate unacceptable levels of anxiety for them, and for this reason they would prefer not to be screened.

Of the 11 (4 per cent) subjects who stated that they did not wish to be tested during pregnancy, six would have accepted screening either preconceptually or in the postnatal period as part of future reproductive decision-making.

For a group of ten (4 per cent), the dilemma of deciding among options raised by a positive test result was the reason why they declined to enter the trial. Of this group six had either a history of infertility or previous pregnancy loss, which they felt contributed to their apprehension at being faced with a major decision regarding continuing a much-wanted pregnancy.

Table 2. Socio-demographic data

	Accepted screening $n=1798$	Declined screening $n=260$
Age (years)		
Mean	28·07 (5·56)	27·74 (5·14)
Range	16–44	16–41
Parity		
0+0	720 (40%)	85 (33%)
0+1>	196 (11%)	30 (12%)
1+0>	882 (49%)	145 (56%)
Gestation (weeks)		
Mean	12·25 (2·12)	12·72 (3·63)
Range	6–18	7–18
Marital status		
Married	1316 (73%)	199 (76%)
Single	409 (23%)	49 (19%)
Divorced	46 (3%)	7 (3%)
Separated	24 (1%)	5 (2%)
Widowed	3 (0·2%)	—
Social class		
1	227 (13%)	35 (13%)
2	553 (31%)	80 (31%)
3	601 (33%)	73 (28%)
4	158 (9%)	25 (10%)
5	91 (5%)	14 (5%)
Unemployed	141 (8%)	22 (8%)
Student	27 (2%)	11 (4%)
Religion		
Protestant	997 (55%)	130 (50%)
RC	243 (14%)	50 (19%)
Christian	47 (3%)	13 (5%)
Other	42 (2%)	15 (6%)
None	469 (26%)	52 (20%)
AFP screening		
Accepted	1776 (99%)	142 (55%)
Declined	22 (1%)	118 (45%)

Nine (3 per cent) simply 'did not want to know', and a further five (2 per cent) stated that they had no reason for refusing, but just did not wish to be screened. Only one woman who declined the test did so because of advances in treatment for CF and ongoing research.

Of the 46 women who were initially undecided, but subsequently refused the test, ten wished more information about the implications of screening. In this

Table 3. Themes of those who declined CF carrier test (percentages in parentheses)

Theme	Declined	Undecided, subsequently declined	Total declined
Against termination of pregnancy	97	14	111 (43%)
Against termination for CF	26	3	29 (11%)
For reasons of partner	17	9	26 (10%)
Consider risk of CF child low	15	4	19 (7%)
Error rate test unacceptable	14	1	15 (6%)
Test causes anxiety or worry	11	2	13 (5%)
Don't want test during pregnancy	11	0	11 (4%)
Require more information	1	10	11 (4%)
Too difficult decision if test positive	8	2	10 (4%)
Don't want to know	9	0	9 (3%)
No reason	5	0	5 (2%)
Advances in treatment or research CF	0	1	1 (1%)
Total	214	46	260 (100%)

group, four wished to discuss the test further with their partner before making a decision, but made no further contact with the clinic, while six felt that they were unlikely to terminate the pregnancy and on this basis declined.

DISCUSSION

Unquestionably, women's opinion on termination of pregnancy was the factor which motivated most to decline the test. This is a similar finding to previous studies (Davies and Doran, 1982).

Of the 111 women who were completely against termination of pregnancy, 78 (70 per cent) had declined AFP screening. In contrast, only six of the 29 (21 per cent) against termination specifically for CF declined AFP screening. It is interesting that 33 (30 per cent) of those who stated that they were completely against termination of pregnancy were planning to participate in the AFP screening programme. One reason for this may be that no reference is made to termination of pregnancy in the hospital's leaflet describing this test, whereas our own CF carrier screening leaflet particularly draws attention to this as a possible consequence. It is noteworthy that two CF carriers identified in the trial subsequently declined AFP screening. This may have been directly due to the anxiety experienced while awaiting their partner's carrier test result, but could arguably be a result of the in-depth counselling carried out in the trial which encouraged couples to address their attitude toward termination of pregnancy.

Prenatal CF carrier screening introduced a new concept of screening in its involvement of the male partner. Some women were reluctant to admit a stable relationship with their partner for fear of jeopardizing their entitlement to state financial benefits. Many made it clear, however, that their partner would be available for screening if required. Twenty-eight per cent of male partners did not read the prescreening information leaflet, a figure comparable to the partners of

Table 4. Examples of quotations from women who declined CF carrier screening

Completely against termination of pregnancy
'For us, killing our unborn child because of any disability is not an acceptable option. I know of no other reason why the test would be helpful, so I wouldn't have it.'
Against termination specifically for CF
'I don't think in these circumstances I would find termination acceptable, so I'd prefer not to start on this route.'
For reasons of partner
'My husband would be far too busy to have the test if it became necessary.'
'It's my partner—he doesn't really know about the pregnancy and he says he hasn't got anything like that—he says it will take some time for him to think about it.'
Consider risk of having a child with CF is low
'As I already have a completely healthy child I don't think I am at high risk of having a child with CF. Also there is no family history.'
Don't want to know
'I would prefer not to know.'
Test causes anxiety or worry
'The worry of finding out I have a single gene until my partner is tested. If he is negative, the worry is unnecessary. If he is positive, even more worry would result until the prenatal diagnosis when if the baby is negative, again the worry has been unnecessary.'
Error rate of the test unacceptable
'As only 85 per cent of cases tested are successful, if my test was negative I'm still going to be anxious that I might be that 15 per cent and as you state you cannot guarantee the child will not have CF.'
Require more information
'Want to know more about the tests that are carried out before making a decision.'
Don't want test during pregnancy
'I feel it is now too late for the test to be relevant. If the test was available for someone intending to become pregnant but not yet pregnant I would have had it at that stage.'
Too difficult a decision if test positive
'I think I would face a very difficult moral dilemma if I discovered, whilst pregnant, that both my husband and I were CF carriers. I would then want to have the baby screened and if it had CF I would be very worried about making a decision to have an abortion, which in theory I'm opposed to, but realistically, don't know what I'd do.'
'My husband and I have decided against the test because we have waited a while for this baby. To find out something was wrong would be shattering for us both, we would rather take our chances and hope everything will be okay.'
No reason
'Don't know—just don't want to have test done.'
Because of advances in treatment for CF
'I would not terminate a pregnancy if CF was diagnosed. Having worked with cystic children and adolescents, medical advances are being made which increase both the quality of life and the life expectancy of the CF sufferer.'

women who accepted the test. This may reflect the findings of previous work, which suggests that it is a minority of men who feel that the couple should together decide about prenatal diagnosis (Sjogren, 1992).

Perhaps not surprisingly, multiparous women were inclined to feel their risk of having a child with CF was low. As one subject stated, 'I have five healthy children already and feel no need for this test.' Genetic conditions are frequently equated with a family history and despite the prescreening leaflet emphasizing that a family history was not a prior condition to being a CF carrier, this was quite a difficult concept for some to comprehend. A previous study has shown that perceived risk rather than actual risk influences women's uptake of prenatal screening tests (Marteau *et al.*, 1991).

In some instances, satisfaction with the care given in previous pregnancies was a factor: 'happy with my last two pregnancies and will just have the usual tests'.

The incompleteness of CF carrier testing was not a major concern and when it was cited as a reason for declining the test, it was frequently connected with concern: 'With the test only being 85 per cent accurate I do not feel it is worth putting myself through the worry.' Nine (69 per cent) of those who declined the CF test on the basis that it would provoke unacceptable levels of stress did accept AFP screening. This could lead one to conclude that it was the false-negative rate of the CF test which likewise deterred most of this anxious group. Data from this trial on the psychological effects of identifying women as CF carriers during pregnancy has been reported (Mennie *et al.*, 1993).

Only 11 (4 per cent) of women actually stated that they were against being screened during pregnancy. 'Feel it is too late to be having the test. Before pregnancy I would have wanted it, including tests for other genetic diseases.' A study of attitudes of recent parents to CF carrier testing found that approximately half of the sample were in favour of screening in early pregnancy (Green, 1992). In this trial, women who have stated a preference to being screened outside pregnancy are invited to request the test in the immediate postnatal period.

A recurring question amongst those who requested further information was the availability of the test if termination of pregnancy was not considered an option or induced feelings of uncertainty. Prenatal diagnosis for those who would not consider abortion has generated argument (Clark and DeVore, 1989; Thorp and Bowes, 1989) and many obstetricians are reluctant to carry out a risk-associated procedure in such circumstances. Nevertheless, 'to be prepared' is a reason that many women cite for wishing to be screened for CF carrier status, and collectively this group constitutes a substantial prescreening counselling component. Certainly, couples where both partners test positive can be given advance warning that their child will be at a 1 in 4 risk of having the disease and can opt not to pursue prenatal diagnosis. As one woman commented, 'it's a non-invasive test and there are a number of points where one can decide whether or not to proceed further'.

In Britain, it is believed that more than one in ten couples experience difficulty in either achieving a pregnancy or having a live-born child (Page, 1988). Studies indicate that couples who have undergone infertility investigations and subsequently encounter prenatal diagnostic procedures experience elements of the psychological trauma associated with their infertility (Sandelowski *et al.*, 1991).

The CF screening test is offered early in pregnancy, at the woman's first antenatal clinic visit. It is understandable that having only just achieved a pregnancy, the prospect of being confronted with a decision to continue or end it is a situation that these couples would prefer to avoid.

The importance of good prescreening information cannot be overemphasized. Studies indicate that informed decision-making requires time (Lorenz *et al.*, 1985). The aim of the prescreening leaflet in this trial was to present a global view of CF carrier screening to women and their partners well in advance of screening, in order that they might make the correct decision with regard to accepting or declining the test. Few came to the clinic undecided; nonetheless, frequent evaluation of the content and delivery of prescreening information and counselling should be part of the on-going evaluation of any prenatal screening programme.

ACKNOWLEDGEMENTS

We are grateful to all the antenatal patients who responded to the questionnaire and took the time to comment. We thank the medical and nursing staff of the Simpson Memorial Maternity Pavilion for making this trial possible. The study was funded by grants from the Cystic Fibrosis Trust and the Scottish Home and Health Department.

REFERENCES

Clark, S.L., DeVore, G.R. (1989). Prenatal diagnosis for couples who would not consider abortion, *Obstet. Gynecol.*, **73,** 1035–1037.

Davies, B.L., Doran, T.A. (1982). Factors in a woman's decision to undergo genetic amniocentesis for advanced maternal age, *Nurs. Res.*, **31,** 56–59.

Field, P.A., Morse, J.M. (1990). *Nursing Research: the Application of Qualitative Approaches*, London: Chapman and Hall, 102–103.

Green, J.M. (1992). Principles and practicalities of carrier screening: attitudes of recent parents, *J. Med. Genet.*, **29,** 313–319.

Lorenz, R.P., Botti, J.J., Schmidt, C.H., Ladda, R.L. (1985). Encouraging patients to undergo prenatal genetic counselling before the day of amniocentesis, *J. Reprod. Med.*, **30,** 933–935.

Marteau, T.M., Kidd, J., Cook, R., Michie, S., Johnston, M., Slack, J., Shaw, R.W. (1991). Perceived risk not actual risk predicts uptake of amniocentesis, *Br. J. Obstet. Gynaecol.*, **98,** 282–286.

Mennie, M.E., Liston, W.A., Brock, D.J.H. (1992a). Prenatal cystic fibrosis carrier testing: designing an information leaflet to meet the specific needs of the target population, *J. Med. Genet.*, **29,** 308–312.

Mennie, M.E., Gilfillan, A., Compton, M., Curtis, L., Liston, W.A., Pullen, I., Whyte, D.A., Brock, D.J.H. (1992b). Prenatal screening for cystic fibrosis, *Lancet*, **340,** 214–216.

Mennie, M.E., Compton, M.E., Gilfillan, A., Liston, W.A., Pullen, I., Whyte, D.A., Brock, D.J.H. (1993). Prenatal screening for cystic fibrosis: psychological effects on carriers and their partners. Submitted.

Page, H. (1988). The increasing demand for fertility treatment, *Health Trends*, **4:20:** 115–118.

Sandelowski, M., Harris, B.G., Holditch-Davis, D. (1991). Amniocentesis in the context of infertility. *Health Care Women Int.*, **12,** 167–178.

Sjogren, B. (1992). Future use and development of prenatal diagnosis. Consumers' attitudes, *Prenat. Diagn.*, **12,** 1–8.

Thorp, J.M., Jr, Bowes, W.A., Jr (1989). Prenatal diagnosis for couples who would not consider abortion (Letter), *Obstet. Gynecol.*, **74,** 828–829.

10. Kirkman R, Chantler E. Contraception and the prevention of STD. *Br Med Bull* (in press).
11. Spieler JM (moderator). Summary discussion. In: Alexander NJ, Gabelnick HL, Spieler JM, eds. Heterosexual transmission of AIDS: proceedings of the Second Contraceptive Research and Development (CONRAD) Program International Workshop, February, 1989. New York: Wiley-Liss, 1990: 419–26.
12. Potts M, Feldblum PJ. Changing behavior: barrier methods in high-risk populations. In: Alexander NJ, Gabelnick HL, Spieler JM, eds. Heterosexual transmission of AIDS: proceedings of the Second Contraceptive Research and Development (CONRAD) Program International Workshop, February, 1989. New York: Wiley-Liss, 1990: 69–79.
13. Chantler E. Vaginal spermicides: some current concerns. *Br J Fam Plan* 1992; **17:** 118–19.
14. Chantler E. New and existing spermicides with virucidal properties. In: Alexander NJ, Gabelnick HL, Spieler JM, eds. Heterosexual transmission of AIDS: proceedings of the Second Contraceptive Research and Development (CONRAD) Program International Workshop, February, 1989. New York: Wiley-Liss, 1990: 303–10.

Negative investigations

Abdominal pain, headache, tiredness, and chest pain are common complaints[1] that are sometimes troublesome enough to prompt a visit to the family physician. A small proportion of patients will be referred for further investigation and treatment. As people prosper they expect to feel healthier and to have good health care, so they are more inclined to consult and to demand specialist attention.[2] Ultimately, there is a rise in both the absolute numbers and the proportion of patients attending clinics with no diagnosis. If outpatient facilities are to be inundated with patients who have no organic disease but who demand to know what is wrong, we need to know whether investigating them is worthwhile. Doing tests is generally presumed to be beneficial. The results may confirm or refute a diagnosis, allow treatment to be targeted and monitored, and reassure both doctor and patient that there is no serious disease. Ordering a test may sometimes enhance the doctor's income and protect against litigation.[3] On the negative side it may encourage illness behaviour, reinforce a belief that there is something to find, and ultimately lead to crippling somatisation.[4]

The decision to conduct an investigation to look for disease is influenced by the seriousness and treatability of the disease as well as by the likelihood of finding it. A serious disease is worth diagnosing even when it is not treatable, because of the need to give a prognosis. This is not always the case for benign untreatable diseases; and for some benign treatable conditions such as peptic ulcer and reflux oesophagitis empirical treatment is usually safe. One could make a case for not investigating dyspepsia with endoscopy in patients aged less than 50 because empirical therapy is safe, effective, and cheap and because the likelihood of missing a treatable cancer is very small.

Occasionally a test is done with the specific purpose of reassuring the patient that nothing is wrong. When used in this way the investigation can be regarded as part of treatment. A benign or possibly irrelevant diagnosis such as hiatus hernia or diverticular disease may be more reassuring than no diagnosis; a peg for the symptoms may aid attribution and legitimise the complaints by giving the patient something to tell relatives and friends. In a study of oesophageal function in patients with angiography-negative chest pain, those who accepted that their pain was related to oesophageal disorder did better than those who did not.[5] This finding suggests that concern about the chest pain is more important than the abnormality that is supposed to underly it. The important question arising from this study is whether the oesophageal tests were necessary for correct attribution and the resulting wellbeing.

Some patients will not be convinced that there is nothing treatable or seriously wrong until a test is done, but many will accept a simple explanation and reassurance for their symptom. Favourable outcome for patients with headache presenting to a neurology clinic was associated with satisfaction with consultation and not with the extent of investigation.[1,6] For those who are difficult to reassure, with or without investigation, there is an alternative to more opinions or tests. Klimes et al,[7] in a randomised trial of a "difficult" group of patients with non-cardiac chest pain, who were not getting any better despite negative test results and reassurance, showed that successful reattribution of symptoms and reduced suffering can be achieved with a cognitive-behavioural intervention programme.

Doctors need to reassess their priorities when investigating patients and to ask themselves three questions. Will a positive investigation really make a difference to my management of this patient? Will a negative investigation help me reassure the patient and help the patient attribute the symptoms to a benign disorder? Is there an alternative, cheaper strategy for managing the patient? The doctor is well equipped to deal with the first question but not the other two. Management trials in which patients are randomly assigned to conventional treatment with and without investigation will help answer the second question. A further intervention arm such as an education and reassurance package will help answer the third. Economic endpoints such as time lost from work, further consultation, and cost of the intervention, as well as symptoms, would also need to be included.

1. Creed F, Mayou R, Hopkins A. Medical symptoms not explained by organic disease. London: Royal College of Psychiatrists and Royal College of Physicians, 1992.
2. Valori RM. Nutcracker, neurosis or sampling bias? *Gut* 1990; **31:** 736–37.
3. Kastor JA. Pacemaker mania. *N Engl J Med* 1988; **318:** 182–83.
4. Bass C, Murphy M. The chronic somatizer and the government white paper. *J R Soc Med* 1990; **83:** 203–05.
5. Ward BW, Wu WC, Richter JE, Hackshaw BT, Castell DO. Long-term follow-up of symptomatic status of patients with non-cardiac chest pain: is diagnosis of esophageal etiology helpful? *Am J Gastroenterol* 1987; **82:** 215–18.
6. Fitzpatrick RM, Hopkins A. Referrals to neurologists for headaches not due to structural disease. *J Neurol Neurosurg Psychiatry* 1981; **44:** 1061–67.
7. Klimes I, Mayou RA, Pearce MJ, Coles L, Fagg JR. Psychological treatment for atypical non-cardiac chest pain: a controlled evaluation. *Pychol Med* 1990; **20:** 605–11.

PUBLIC HEALTH

Prenatal screening for cystic fibrosis

MOIRA E. MENNIE ANNETTE GILFILLAN MARY COMPTON
LUCY CURTIS W. A. LISTON I. PULLEN DOROTHY A. WHYTE
D. J. H. BROCK

Screening for carriers of CF (cystic fibrosis) is now possible but the best way of delivering such a service is unknown. In one model 4348 women attending antenatal clinics in an Edinburgh maternity hospital were invited to participate in a trial of prenatal screening. Mouthwash samples were tested for six CF alleles (85% of mutant genes) and when a woman was found to be a CF carrier her partner was also tested. Heterozygous couples were offered prenatal diagnosis.

609 (14%) women declined to enter the trial and another 574 (13%) were not screened, usually because of late booking. Among the remaining 3165 women there were 111 carriers of a CF gene (1 in 29). 4 of these 111 had carrier partners and these couples opted for prenatal diagnosis, the 1 pregnancy with an affected fetus being terminated. The psychological impact of screening was assessed by the general health questionnaire. There was a significant increase in stress at the time of the test result among women identified as carriers. However, this disappeared when their male partners tested normal and did not reappear later in the pregnancy.

By providing time for couples to discuss the possibility of screening and by offering the test at a point (the antenatal booking clinic) at which most pregnant women are seen, this approach has advantages, provided that counselling is readily available.

Lancet 1992; **340**: 214–16.

Introduction

The cloning of the cystic fibrosis (CF) gene in 1989[1-3] and the demonstration that 70% or so of mutations are the ΔF508 allele,[4] made it possible to contemplate prenatal screening. In the UK the Cystic Fibrosis Research Trust solicited bids for trial projects aimed at delivering screening either through community health services (family-planning clinics or general practitioners) or in antenatal clinics. Although there are some advantages in focusing testing on individuals or couples before conception experience in screening for other autosomal recessive genetic disorders suggests that testing during pregnancy is more effective.[5]

In our trial project we assessed screening in the antenatal clinic of Edinburgh's largest maternity hospital, the Simpson Memorial Maternity Pavilion, which has about 5000 deliveries a year. Although couple screening has recently been proposed[6] we used the more conventional two-step model. Women were offered testing at their first antenatal clinic visit; if they were negative no further action was taken but if they were positive the partner was tested. When both parents carried CF alleles—ie, there was a 1-in-4 risk of an affected child—prenatal diagnosis was offered. If the father was negative the residual risk was explained to the couple in a counselling session, but no further action was taken.

The difficulty with this and any other method of screening lies in the molecular heterogeneity of CF. Over one hundred and fifty mutant CF alleles have been described (International CF Genetics Analysis Consortium, personal communication), many very rare. It is only possible to test for the more prevalent mutations in a specific population and to calculate residual risks on the basis of known allele frequencies. We have looked for six mutations representing some 85% of those found in Scotland.[7] Residual risks are outlined in fig 1.

Methods

Recruitment

Before the full trial 180 women were sent an information leaflet and questionnaire to test their reactions.[8] On the basis of the responses received (81%), a printed leaflet was designed describing the main features of CF and the methodology of the screening trial. This leaflet was sent to all women with their booking clinic appointment; they were asked to discuss it with their partners and were invited to join the trial by signing a consent form. The leaflet emphasised that screening is imperfect; it reduces but does not abolish the risk of an affected child. Women were advised not to participate if they could not identify the baby's father. Other exclusions are shown in the table.

Counselling

At the clinic the midwife responsible for booking asked the patient whether she had read the leaflet, understood it, and wished to join the trial. Women who had not read the leaflet (often because of reading difficulties) or found it too complex were counselled with visual aids by a genetics nurse. Women carrying a CF allele were

ADDRESSES: **Human Genetics Unit** (M. E. Mennie, HV, A. Gilfillan, BSc, M. Compton, RM, L. Curtis, BSc, Prof D. J. H. Brock, PhD); **and Departments of Obstetrics and Gynaecology** (W. A. Liston, MB); **Psychiatry** (I. M. Pullen, MB); **and Nursing Studies** (D. A. Whyte, PhD), **University of Edinburgh, Edinburgh, UK.** Correspondence to Prof D. J. H. Brock, Human Genetics Unit, Western General Hospital, Edinburgh EH4 2XU, UK.

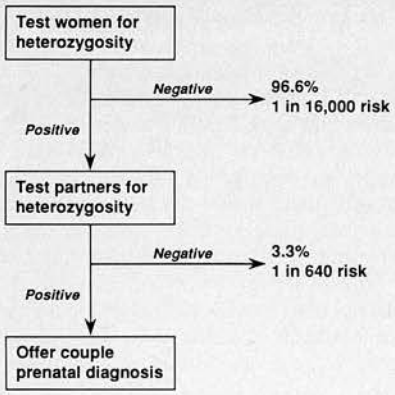


Fig 1–Two-step screening.

later seen with their partners for further counselling by the nurse. If both had CF alleles they were referred to a consultant obstetrician for discussion of possible prenatal diagnosis.

Psychological status

All women entering the trial were asked to complete a twelve-item general health questionnaire (GHQ)[9] before testing (threshold GHQ). Carriers filled in a GHQ at the time of their test result (GHQ1), at the time of their partner's test result (GHQ2), and again 6 weeks later (GHQ3). For every carrier 2 control women of the same parity who had received negative results were selected from the same clinic and tested at comparable times.

Laboratory analyses

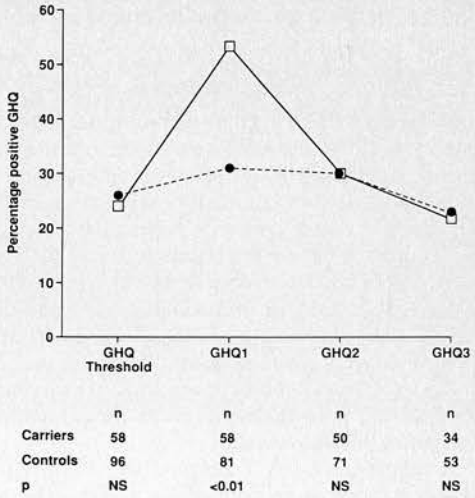
In the early part of the trial blood samples were collected from women and mouthwash samples from their partners. However, mouthwash samples had fewer difficulties and all participants were subsequently asked to rinse their mouths briefly with 10–15 ml tap-water which was transferred to a Universal container. Buccal cells were pelleted by centrifugation, suspended in 50 mmol/l sodium hydroxide, heated in a boiling water bath for 20 min, neutralised, and centrifuged.

Two assay systems were used, with overlap in the mutant alleles detected. In the in-house assay[7] exons 10 and 11 were simultaneously amplified by the polymerase chain reaction; the ΔF508 and ΔI507 alleles in exon 10 were detected by electrophoresis on polyacrylamide gels, whereas the G551D and R553X alleles in exon 11 were detected by differential restriction enzyme digestion. The commercial assay (courtesy of Cellmark)[10] used the amplification refractory mutation system (ARMS) to detect ΔF508, G551D, G542X, and 621 + 1G→T. The in-house assay detected 79% and the ARMS system 83% of mutations; in combination they allowed scanning for 85% of mutations.

DELIVERY OF PRENATAL SCREENING (OCTOBER, 1990, TO APRIL, 1992)

Women offered screening	4348
Declined offer	609 *(14%)*
Not screened	
Late gestation (>18 wk)	430
Abnormal pregnancy	73
Partner unavailable	56
Other*	15
Screened	3165 *(73%)*
Carriers	
ΔF508	97
G542X	7
G551D	6
621 + 1G→T	1
ΔI507	0
R553X	0
Carrier couples	4

*12 women from ethnic groups where incidence of CF is low and whose command of English was inadequate; 1 with a triplet pregnancy; 1 pregnant by artificial insemination from a donor already screened for CF alleles; and 1 not pregnant.



	n	n	n	n
Carriers	58	58	50	34
Controls	96	81	71	53
p	NS	<0.01	NS	NS

Fig 2—GHQ results.

Percentages of carriers (□) and controls (●) with positive responses (3+) shown for each test period.
Numbers of carriers and controls tested shown below figure.

Reporting of results

Participants were told that initial testing would take 7 days and that at that time they could assume that no mutant alleles had been detected. Women who tested positive were informed by telephone if possible or by letter otherwise and an appointment was made for counselling, together with their partner. Partner's samples were tested as quickly as possible and the results communicated by telephone. All results were recorded in the obstetric notes; positive results were also communicated by letter to the woman, her general practitioner, and the consultant obstetrician.

Results

Screening was introduced in October, 1990, the offer being made initially in just one of nine weekly antenatal clinics. By April, 1992, 4348 women in 431 clinics had received the information leaflet and an invitation to participate (table). Most of the 609 women who refused to take part gave their reason as non-acceptance of the possibility of termination in the event of an affected fetus. A further 574 women were not screened because they were already over 18 weeks' gestation (430), because of abnormal pregnancies (mainly blighted ova) (73), or because their partner would not participate (56). Other reasons are shown in the table.

Among the 3165 women tested there were 111 CF heterozygotes, giving a carrier rate of 1 in 29. Since the detection rate of CF alleles was 85%, the true heterozygote frequency is 1 in 24. Partners of 110 of these 111 carriers were screened and 4 (3 ΔF508, 1 G551D) were found to be positive. All 4 of these couples opted for prenatal diagnosis, 3 via amniocentesis and 1 via a transabdominal chorionic villus biopsy. 1 woman was found to be carrying a ΔF508 homozygote and she decided on termination; the diagnosis was confirmed on fetal tissues. The other 3 women were carrying unaffected fetuses and are proceeding to term.

GHQs were regarded as positive if the score was 3 or more.[11] At the time of the test result (GHQ1), 53% (95% confidence interval [CI] 41–66%) of carriers had positive results as against 31% (95% CI 21–41%) of controls

($p < 0{\cdot}01$). This difference had disappeared by the time of the partner's negative test result (GHQ2) (fig 2).

Discussion

The cloning of the CF gene prompted considerable discussion about population heterozygote screening and the consensus was that trial projects of different modes of delivery, not necessarily mutually exclusive, were needed. One method is preconception screening, which provides carrier couples with several options (such as changing partners, artificial insemination by a screened donor, forgoing reproduction, or prenatal diagnosis) and time for reflection and an unpressured choice. However, it is difficult to see how delivery through general practices and family-planning clinics could reach a broad range of the population, and bias towards take-up by the educationally and socially advantaged seems unavoidable.

CF heterozygosity is of medical significance when a pregnancy is planned or in progress, so it makes sense to target screening close to pregnancy. Evidence from other recessively inherited disorders suggests that screening during pregnancy is the most practicable time. In a 1989 US report on screening for Tay-Sachs disease, in which participants could be tested before or during pregnancy, 80% of respondents were already pregnant when the test was sought.[13] In the UK almost all pregnant women now attend hospital at some stage and the antenatal clinic is an effective way of ensuring that screening is offered to as many woman as possible. Reducing the number of delivery points would be of great importance if screening for CF is to become routine, because of the need for back-up counselling at every point.

In the two-step model of delivery tested here, the take-up rate was high (86%) but because of late booking and other reasons the proportion of women screened was 73%. This may represent an effective upper limit for this type of programme. An earlier study showed that some 40% of women decided to enter the programme without reference to their partners[8] but we found that only 1 of the 111 female carriers was unable (or unwilling) to persuade her partner to be tested. This suggests that the motivation of the woman is an important contributor to a good take-up rate—and that non-paternity[14] is not likely to be an important source of error. We have also noted that first-degree relatives of 15 of the 111 carriers detected by screening have made appointments with the genetic counselling clinic to establish their carrier status.

One concern in this trial was stress in women identified as carriers. 20–30% of women had positive GHQ scores before they were tested, a result which accords with previous findings of 35% in pregnancy.[15] Women identified as carriers were significantly more likely to record a positive GHQ than controls but this difference disappeared once their partners had been tested and found not to carry a detectable CF allele. Thus stress would appear to be of short duration in carrier women, despite their being warned that their residual risk of bearing a CF child was still 1 in 600 or so.

The model of prenatal screening for CF tested here has considerable merit. Information leaflets about CF and screening can be studied by women and their partners at home, since couples have 4 weeks or so before any decisions need to be made. Most of their questions can be answered by midwives during the booking-in procedure. More than 96% of screened women had no detectable CF alleles in our series and there is no evidence for an increase in stress amongst this negative group. For an allele detection rate of 85% only 1 in every 865 couples will need to be referrred to a doctor for detailed discussion of prenatal diagnosis.

If about 73% of women offered testing are screened and if the detection rate for CF alleles continues at 85% we would expect to identify about half ($0{\cdot}73 \times 0{\cdot}85 \times 0{\cdot}85$) the 1-in-4 risk couples in our population. We do not know what proportion of these would opt for prenatal diagnosis and termination of pregnancy if the fetus were affected; all 4 of our couples indicated that this was their intention. However, even if only half the parents of an affected fetus chose termination, this would still lead to the reduction in live-born incidence of 1 case of CF for every 10 000 tests. The Royal College of Physicians[16] estimates the cost of treating a CF child at least £5000 per year (1986 figures); over an expected life-span of 25 years this represents a sum of £125 000. Laboratory costs for 10 000 tests are about £30 000, while the total programme costs (including laboratory) are about £80 000. Thus a crude cost-benefit calculation suggests that screening of this kind represents good value. The major resource implicated is in the management of carrier women in the period between their test result and that of their partner. At least one counselling session is needed to answer questions, allay fears, and place risks in perspective. This can be done by informed non-medical personnel with good communication and counselling skills. In our experience a specialist genetics nurse is essential to the smooth delivery of this type of programme.

We thank the medical and nursing staff of the Simpson Memorial Maternity Pavilion for making this trial possible. The study was funded by grants from the Cystic Fibrosis Research Trust and the Scottish Home and Health Department.

REFERENCES

1. Rommens JM, Iannuzzi MC, Kerem B, et al. Identification of the cystic fibrosis gene: chromosome walking and jumping. *Science* 1989; **245:** 1059–65.
2. Riordan JR, Rommens JM, Kerem B, et al. Identification of the cystic fibrosis gene: cloning and characterisation of complementary DNA. *Science* 1989; **245:** 1066–73.
3. Kerem B, Rommens JM, Buchanan JA, et al. Identification of the cystic fibrosis gene: genetic analysis. *Science* 1989; **345:** 1073–80.
4. McIntosh I, Lorenzo ML, Brock DJH. Frequency of F508 mutation on cystic fibrosis chromosomes in UK. *Lancet* 1989; ii: 1404.
5. Modell B. Cystic fibrosis screening and community genetics. *J Med Genet* 1990; **27:** 475–479.
6. Wald NJ. Couple screening for cystic fibrosis. *Lancet* 1991; **338:** 1318–19.
7. Shrimpton AE, McIntosh I, Brock DJH. The incidence of different cystic fibrosis mutations in the Scottish population: effects on prenatal diagnosis and genetic counselling. *J Med Genet* 1991; **28:** 317–321.
8. Mennie ME, Liston WA, Brock DJH. Prenatal cystic fibrosis carrier testing: designing an information leaflet to meet the specific needs of the target population. *J Med Genet* 1992; **29:** 308–312.
9. Goldberg D, Williams P. A user's guide to the general health questionnaire. Windsor: NFER-Nelson, 1988.
10. Ferrie RM, Robertson NH, Vaudin SA, Schwarz M, Little S. Development of multiplex ARMS test for the simultaneous detection of four common CF mutations. *Am J Hum Genet* 1991; **49:** 993A.
11. Mari J, Williams P. A comparison of the validity of two psychiatric screening questionnaires (GHQ12 and SRQ20) in Brazil, using relative operating characteristic analysis. *Psychol Med* 1985; **15:** 651–59.
12. Watson EK, Mayall E, Chaspple J, et al. Screening for carriers of cystic fibrosis through primary health care services. *BMJ* 1991; **303:** 504–7.
13. Shapiro DA, Shapiro LR. Pitfalls in Tay-Sachs carrier detection: physician referral patterns and patient ignorance. *NY State J Med* 1989; **89:** 317–19.
14. Brock DJH, Shrimpton AE. Non-paternity and prenatal genetic screening. *Lancet* 1991; **338:** 1151.
15. Sharp DJ. Validation of the 30-item General Health Questionnaire in early pregnancy. *Psychol Med* 1988; **18:** 503–07.
16. Royal College of Physicians. Prenatal diagnosis and genetic screening. London: RCP, 1989.

Reprinted from Journal of Medical Genetics, 1992; **29**: 5

Prenatal cystic fibrosis carrier testing: designing an information leaflet to meet the specific needs of the target population

M E MENNIE, W A LISTON, D J H BROCK

BMJ Publishing Group, Tavistock Square, London WC1H 9JR

J Med Genet 1992; 29: 308-312

Prenatal cystic fibrosis carrier testing: designing an information leaflet to meet the specific needs of the target population

M E Mennie, W A Liston, D J H Brock

Abstract

A questionnaire was given to 180 patients in an antenatal clinic, who were eligible to enter a pilot trial of cystic fibrosis (CF) carrier testing, seeking their views on the information leaflet inviting them to participate; 161 patients (89%) entered the trial and 145 patients (81%) responded to the questionnaire, including 10 who did not enter the trial. Ninety-six percent of these found the leaflet easy to understand, and 97% of those partners who read the leaflet also found it easy to understand. Fifteen percent of patients thought the leaflet should give additional information. Most (92%) had heard of CF before reading the leaflet, television being the most common source of information. Although avoiding the birth of a child with CF was the reason most patients gave for wishing to be screened, almost as many were interested to know their carrier status. The decision to accept or decline testing was taken in conjunction with their partner by 63% of women. Of those who were screened, 59% stated that taking the test made them feel reassured, while 38% felt slightly apprehensive. It was concluded that, with a number of minor amendments, the leaflet met the specific needs of the target patient population.

The cloning of the cystic fibrosis (CF) gene in 1989 and the subsequent description of a number of mutant CF alleles has made it possible to detect CF carriers in the general population. A survey of Scottish CF patients and their parents shows that about 85% of carriers can be identified by testing for just four mutations.[1] Although this is less than ideal for a screening test, it has been argued that the severity of the disorder demands an immediate start to pilot trials of carrier detection.[2] Accordingly we have initiated such a trial in selected antenatal clinics of the major Edinburgh maternity hospital.

Pregnant women are offered the chance of joining the trial by means of a leaflet sent with their antenatal booking clinic appointment. The leaflet emphasises the incomplete nature of the test and that it cannot guarantee against the birth of a CF child. Volunteer women are screened and if found to carry a CF allele their partners are also screened. Couples where both partners are found to be carriers are offered prenatal diagnosis.

A prerequisite to an informed decision to undergo any screening or diagnostic test is knowledge of the test.[3] Indeed, some argue that the effectiveness of such a screening programme should be assessed by whether the participants have become fully informed.[4] However, to be effective and useful, educational materials must meet the specific needs of the target population.[5]

The use of a questionnaire to assess patients' attitudes to information leaflets has been described.[6,7] Before committing ourselves to a final version of the information leaflet designed for this trial, we tested the acceptability and usefulness of a preliminary version on our target population, by means of a self administered questionnaire.

Subjects and methods

A pilot trial was run in one antenatal booking clinic for a period of 14 weeks. Along with their booking clinic appointment, patients received a copy of the information leaflet outlining the purpose and process of prenatal CF carrier screening (table 1). The leaflet also incorporated a consent form to be signed by patients entering the screening trial.

A total of 200 antenatal patients attended the booking clinic during the pilot trial. Twenty of these patients were not eligible to enter the screening trial for reasons of late gestation (greater than 18 weeks), abnormality of pregnancy (for example, blighted ovum), or unavailability of partner. Questionnaires were not issued to this group. The remaining 180 antenatal patients were given a questionnaire and a stamped addressed envelope at the booking clinic and asked to complete it at home and return it. Patients were not asked to identify themselves. A total of 145 (81%) returned their questionnaire. Of the 161 patients who entered the trial, 135 responded. Of the 19 patients declining to enter, 10 responded.

Results

PATIENTS' PREVIOUS KNOWLEDGE OF CF AND SOURCE OF INFORMATION

The questions were:

(1) Had you heard of the disease cystic fibrosis before you read the leaflet?

Human Genetics Unit, Department of Medicine, University of Edinburgh, Western General Hospital, Edinburgh EH4 2XU.
M E Mennie
D J H Brock

Simpson Memorial Maternity Pavilion, Edinburgh.
W A Liston

Correspondence to Dr Mennie.

Received 28 May 1991.
Revised version accepted 6 September 1991.

Table 1 The information leaflet.

Cystic fibrosis carrier testing
Prenatal screening trial

One in every 25 men and women carry a *single* cystic fibrosis gene. A single cystic fibrosis gene is completely harmless. But, if a woman who carries a single cystic fibrosis gene has a child by a man who also carries a single cystic fibrosis gene, there is a *1 in 4 chance* that their child will have the disease *cystic fibrosis*.

(diagram of cystic fibrosis genetics)

Cystic fibrosis

Cystic fibrosis is serious. The average life expectancy is 19 years. Children with cystic fibrosis have repeated chest infections and are usually underweight and undersize. They may need daily physiotherapy and large numbers of tablets to assist digestion. If they live long enough they will probably need a heart and lung transplant.

We think that a woman who is having a baby will want to know the answers to three important questions:

(1) Am I a carrier?
(2) Is my partner a carrier?
(3) Will my baby be affected?

We are running a trial which tests whether or not you are a carrier of a single cystic fibrosis gene. If you are *not* there is no need for further concern.

If you *are* a carrier it is possible to tell if your husband or partner is also a carrier.

Only if *both* of you are carriers will you have a 1 in 4 chance of having an affected child.

If you and your partner are *both* carriers, a prenatal test can tell whether or not your baby will be affected.

Prenatal diagnosis

This is *one* option available to couples who have a 1 in 4 chance of having a child with cystic fibrosis. It is possible to diagnose cystic fibrosis in the fetus. If the prenatal diagnostic test shows that the unborn child is going to have cystic fibrosis, you may wish to have an abortion.

Prenatal diagnosis is *not* recommended for those couples who find abortion unacceptable.

The cystic fibrosis carrier screening test

A small sample of the blood taken for your other antenatal tests will be used to test if you are a carrier. You will be contacted *only* if the test shows you are a carrier.

Testing for the cystic fibrosis gene is still only possible in *85%* of cases. This means we cannot guarantee that you will *not* have a child with cystic fibrosis. However, if the test is negative it will greatly reduce that risk.

Testing your partner

If you are found to carry a single cystic fibrosis gene we would be able to test your partner.

A small mouth wash sample will be used to test your partner, so he will *not* have to give a blood sample.

If your partner's test is negative then your risk of having a baby with cystic fibrosis is *low*.

A special leaflet and genetic counselling is available for all couples where *one* partner has a positive carrier test.

Couples who are both carriers of a single cystic fibrosis gene

Genetic counselling is available to couples where *both* partners are found to be carriers.

The various options open to you will be fully explained. You will have time to *think* through these options and to *discuss* them at length with the counsellor.

Further information and advice about prenatal carrier testing for cystic fibrosis

There will be time at the booking clinic to ask any questions, or discuss any concerns you may have about prenatal carrier testing for cystic fibrosis. Counselling is available at any stage of the screening trial. There will be a nurse counsellor available at the antenatal booking clinic.

Prenatal carrier testing for cystic fibrosis is voluntary

Prenatal carrier testing for cystic fibrosis is *entirely voluntary*.

If you would like to be tested please complete the enclosed form and bring it with you to the antenatal booking clinic.

Important points to remember about prenatal carrier testing for cystic fibrosis

(1) Carrying a single cystic fibrosis gene is completely harmless.
(2) Only if *both* partners carry a single cystic fibrosis gene will there be a 1 in 4 chance that their child will have cystic fibrosis.
(3) Testing for the cystic fibrosis gene is still only possible in *85%* of cases.
(4) Genetic counselling is available for all couples where both partners are found to carry a single cystic fibrosis gene.
(5) The prenatal cystic fibrosis carrier screening trial is entirely voluntary.
(6) Remember there are trained staff to give you information and advice – *please do ask*.

(2) If you had heard of cystic fibrosis, where did you hear about it? (Tick as many options as required.)

There were 145 responses, with 134 (92%) saying yes. Major sources of information were TV, women's journals, and charity appeal.

PATIENTS' ASSESSMENT OF THE LEAFLET AND UNDERSTANDING OF THE PURPOSE OF CF CARRIER TESTING

The questions were:

(3) In your opinion, is the leaflet introducing the cystic fibrosis carrier screening test difficult or easy to understand?
(4) How many times did you read the leaflet before you felt you understood it?
(5) Do you feel you understand the purpose of cystic fibrosis carrier testing?
(6) Do you feel the leaflet should give more information?
(7) Has your partner read the leaflet?
(8) If your partner read the leaflet, was it, in his opinion, difficult or easy to understand?
(9) Did you make the decision to accept or refuse the screening test alone or with your partner?

The answers of the 145 respondents are listed in table 2.

PATIENTS' REASONS FOR BEING SCREENED (NUMBERS BASED ON THOSE 135 RESPONDENTS WHO ENTERED THE SCREENING TRIAL)

The question asked was:

(10) If you have decided to take the screening test, is this because (tick as many options as required)
  (a) I am interested to know if I carry a single cystic fibrosis gene. (96 respondents)
  (b) I do not want my child to have cystic fibrosis. (102 respondents)
  (c) My partner wanted me to have the screening test. (22 respondents)
  (d) Other (please state reason). (10 respondents)

PATIENTS' REASONS FOR NOT BEING SCREENED (NUMBERS BASED ON THE 10 RESPONDENTS WHO DID NOT ENTER THE TRIAL)

The question asked was:

(11) If you have decided not to take the screening test is this because (tick as many options as required)
  (a) I would prefer not to know if I carry a single cystic fibrosis gene. (No respondents)
  (b) I am against termination of pregnancy. (10 respondents)
  (c) My partner did not want me to take the screening test. (3 respondents)
  (d) Other (please state reason). (No respondents)

Table 2 Patients' assessment of information leaflet and their understanding of CF carrier testing: numbers (and percentages).

Assessment of leaflet	Difficult to understand 6 (4)			Easy to understand 139 (96)	
Times leaflet read before understanding	Once 94 (65)	Twice 46 (32)	Thrice 2 (1)	Four times 1 (1)	Many times 2 (1)
Assessment of understanding of the purpose of CF screening	Understand 141 (97)			Don't understand 4 (3)	
Wish for more information in the leaflet	Yes 21 (15)		No 108 (74)	Don't know 16 (11)	
Number of partners who read the leaflet	Yes 92 (63)		No 50 (35)	Don't know 3 (2)	
Partner's assessment of the leaflet	Difficult to understand 3 (3)			Easy to understand 89 (97)	
Partners who participated in the screening decision	Decision taken with partner 91 (63)			Decision taken without partner 54 (37)	

PERSONS WITH WHOM PATIENTS DISCUSSED THE SCREENING TEST (NUMBERS BASED ON RESPONDENTS WHO ENTERED THE TRIAL)

The question asked was:

(12) Have you discussed taking the screening test with anyone? (Tick as many options as required)

(a) Partner	(112 respondents)
(b) Relative	(18 respondents)
(c) Friend	(21 respondents)
(d) GP	(5 respondents)
(e) Health visitor	(1 respondent)
(f) Other (state who)	(no respondents)

PATIENTS' ESTIMATION OF THEIR LIKELIHOOD OF BEING A CF CARRIER AND ATTITUDE TO BEING TESTED (NUMBERS BASED ON THOSE 135 PATIENTS WHO ENTERED THE SCREENING TRIAL)

The questions were:

(13) How likely do you think it is that you will carry a single cystic fibrosis gene?

(14) How does the thought of the cystic fibrosis carrier test make you feel?

Four options were listed (table 3).

Discussion

The cystic fibrosis pilot trial began in the antenatal clinic of the Simpson Memorial Maternity Pavilion in October 1990. Since the first programmes of maternal serum AFP screening were developed and started in Edinburgh in 1975, we already had in place antenatal staff broadly trained in the process of explaining the concepts of screening.

With their antenatal booking clinic appointment, patients received a CF prescreening information leaflet incorporating a consent form. The midwife carrying out the booking procedure ensured that her patient had received a leaflet, had read it (or been able to read it), and was fully aware of the consequences of CF carrier screening. A genetic nurse was on duty at the clinic to answer patient or staff questions and to offer counselling and one to one sessions, using visual aids, to patients and couples with difficulty in reading or understanding the leaflet. Patients wishing to enter the trial signed a consent form.

Information given before prenatal genetic screening must be adapted to a woman's circumstances and be sufficient to enable her to reach a fully informed decision.[8] Only too frequently, when educational protocols are being developed, the target population reaction is not sought.[9] For information and education to be effective, this group needs to be consulted to determine what they need to know or learn. This study attempts to consult antenatal patients about a leaflet designed to inform them about prenatal CF carrier testing.

The response to the questionnaire was high (81%). All questionnaires were completed in full and many respondents commented at length on the questions. Most (92%) claimed they had heard of CF before they read the information leaflet, and many were able to cite the source of their knowledge. This probably reflects the high profile of the Cystic Fibrosis Research Trust in the UK and may not be applicable to other European countries.

There was no correlation between absence of previous information and difficulty understanding the leaflet. Of the six respondents who had difficulty understanding the leaflet (table 2), five had previously heard of CF. However, of the four women who felt they did not understand the purpose of CF screening (table 2), three stated they had not previously heard of CF. Comments indicated that many of this antenatal patient population were also familiar with one major clinical feature of CF namely lung disease. A concern raised regarding the initiating of CF carrier screening programmes is that only a small percentage of the population are at present familiar with the disease.[4] This study did not attempt to test subjects' knowledge of CF and it would be misleading to suggest that patients had detailed knowledge. Indeed, more details about the disease itself were requested by all 15% of respondents (table 2) who felt that the leaflet should give more information. Nevertheless, it is encouraging that so many women were previously aware of the disorder.

The Council of Europe, in their official statement on genetic screening, note the role that the media can play in informing and educating the public and recommend they be

Table 3 Patients' estimation of their likelihood of being a CF carrier and their attitude to being screened: numbers (and percentages).

Estimated likelihood of being a CF carrier	Highly likely 1 (1)	Not very likely 62 (46)	Most unlikely 72 (53)	Don't know 0
Antenatal patient's attitude to being screened	Anxious 4 (3)	Slightly apprehensive 51 (38)	Reassured 79 (59)	Don't know 1 (1)

kept informed of all aspects of antenatal genetic screening.[8] In this study, television was the leading source of information about CF. A considerable number of respondents were acquainted with the disease through women's journals and charity appeals. Knowledge of CF was acquired by a number of women through their occupation and in three cases by virtue of having a distant family history of the disorder.

The majority (96%) of antenatal patients stated that they found the leaflet easy to understand (table 3). There was no correlation between difficulty understanding the leaflet, or failing to understand the purpose of CF screening, and declining the test. All 10 respondents who declined the test found the leaflet easy to understand and stated that they felt they understood the purpose of CF screening. One woman took the CF test, despite finding the leaflet difficult to understand and not understanding the purpose of CF screening.

There is a danger in designing an information leaflet that one may make it either too simple or too complex. Many women are confident and articulate users of the antenatal care system and can cope with detailed information. Others lack the basic knowledge to know the right questions to ask. Our solution has been to design a comparatively simple leaflet and to train midwives to pick up those cases who are unsure about what is being offered. A genetic nurse is on standby to give one to one counselling and answer the more sophisticated questions.

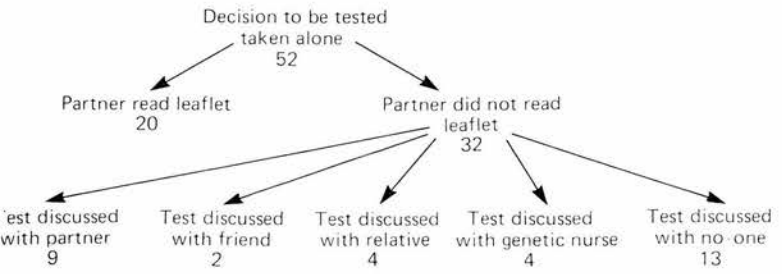
Most women (65%) stated they had only read the leaflet once before feeling they understood it (table 2). Antenatal bookers receive a considerable volume of material to read along with their clinic appointment and may find they have limited time to read and absorb it all. Those who enter the CF screening trial must sign a consent form and in addition the midwife responsible for booking a patient is instructed to do his or her utmost to ensure that a participant understands the purpose of the screening test. Nevertheless, studies do indicate that a lack of knowledge and understanding of women participating in prenatal screening programmes is an area for concern[10] and researchers have attempted to design instruments whereby knowledge of such tests can be measured.[3] CF carrier screening may carry more obscure risks and considerations than other antenatal screening tests, notably perceived change of self image in those identified as gene carriers.[11]

A prerequisite of prenatal CF carrier screening is cooperation of the male partner. Most partners (63%) read the prescreening leaflet and were involved in the test decision (table 2). The partners of all 10 women who declined the test had read the leaflet. Although two of these women took the decision to decline the test alone, they had discussed it with their partner. Of those who found the leaflet difficult to understand, five partners read the leaflet and three also found it difficult to understand. Nevertheless, they were party to the decision to be screened. All partners of those women who felt they did not understand the purpose of CF screening were involved in discussion and decision making, although only one had read the leaflet.

Twenty-one respondents (14%) who entered the trial discussed taking the test with a friend and 18 (12%) with a relative. Only five respondents had talked with their GP and one woman to her health visitor. Fifty-two women made the decision to take the test alone. A concern which emerged from this study is that 13 (9%) women took the decision to be tested without discussing the test with either their partner or other significant person (figure). Evidence suggests that social network and social support play an important role in health, individual decision making, and behaviour. Moreover, social support is known to serve a protective function by moderating the impact of stress and facilitating coping.[12] It could be hypothesised that good social support will contribute positively to a woman's adjustment to being a CF carrier. It has been pointed out that the whole area of psychosocial support has been neglected in the provision of antenatal care and that careful assessment of a woman's social support system should be an integral part of the antenatal booking procedure.[13] Identifying vulnerable women in advance of any screening test is important. This study suggests that around 10% of women booking at the antenatal clinic may have reduced perceived or received social support. Further research is needed to substantiate this.

Those women who decided to accept the CF screening test did so, in 76% of cases, to avoid the birth of a child with CF. In addition, 71% stated, either with or without saying they wished to avoid having a CF child, that they were interested to know if they carried a single CF gene. In 16% of cases, the male partner's wishes had been instrumental in their decision to be tested. Those who gave additional reasons for being screened (7%) stated they wished to help with research or contribute to a better understanding of the disorder. Commendable though this is, we have subsequently discouraged women from entering the trial for these reasons. A positive test result has far reaching consequences which prospective participants must consider carefully.

A vast majority (99%) of women who accepted the screening test thought it 'not very likely' or 'most unlikely' that they would be found to be carriers (table 3). Nevertheless,



nalysis of discussion with significant others, among those women who took the decision lone, to enter the CF screening trial.

41% were still 'anxious' or 'slightly apprehensive' about being screened. There was no correlation between difficulty in understanding the leaflet or understanding the purpose of CF screening and feelings of anxiety towards being tested. The one participant who stated the test made her feel anxious had taken the decision to be tested only after discussion with her GP. She did not think it very likely that she would carry a single CF gene. The whole area of anxiety generated by this programme of screening will be explored in greater depth.

"More about people with the disease, effects, lifespan, etc" was a typical request made by the 15% of respondents who felt that the leaflet should give more information (table 2). However, those who felt the leaflet offered sufficient information frequently commented that, although they would like to know more about the disease, additional information in the leaflet might only serve to confuse. As a compromise we expanded, slightly, the section of the leaflet entitled 'The disease cystic fibrosis' and, as an adjunct, designed a separate leaflet devoted to the disorder. This leaflet is made readily available at the antenatal clinic.

Good quality educational materials can help promote the relationship between the patient and the health care professional as well as enhancing patient knowledge and self care.[5] Quality of text will be determined by its readability and must, therefore, be written on a level that is appropriate for the target population. Moreover, readability will be influenced by the content, style, layout, colour, and illustrations. It is unrealistic to believe that a leaflet can provide blanket coverage of all aspects of antenatal CF carrier screening. A mixture of information giving, teaching, and counselling is needed. Nevertheless, the delivery of infor mation by leaflet is one very importan strategy. We shall, therefore, continue t evaluate the delivery of prescreening informa tion by leaflet for the duration of this trial.

We are grateful to all the antenatal patient who responded to the questionnaire and too time to comment about the CF carrier testin prescreening information leaflet. Copies of th leaflet may be obtained from the authors o request. This work was supported by a gran from the Cystic Fibrosis Research Trust.

1 Shrimpton AE, McIntosh I, Brock DJH. The incidence c different cystic fibrosis mutations in the Scottish popul tion: effects on prenatal diagnosis and genetic counsellin *J Med Genet* 1991;**28**:317–21.
2 Brock DJH. Population screening for cystic fibrosis. *Am Hum Genet* 1990;**47**:164–5.
3 Marteau TM, Johnston M, Plenicar M, *et al*. Developmer of a self-administered questionnaire to measure women knowledge of prenatal screening and diagnostic tests. *Psychosom Res* 1988;**32**:403–8.
4 Wilfond BS, Fost N. The cystic fibrosis gene: medical an social implications for heterozygote detection. *JAM* 1990;**263**:2777–83.
5 Farrell-Miller P, Gentry P. How effective are your patie education materials? Guidelines for developing an evaluating written educational materials. *Diabetes Edt* 1989;**15**:418–22.
6 Bhopal RS, Gilmour WH, Fallon CW, *et al*. Evaluation of practice information leaflet. *Fam Pract* 1990;**7**:132–7.
7 Merrill CR, Knox AM. Patient attitudes to body compute tomography: use of an information leaflet. *Clin Radi* 1986;**37**:537–8.
8 Council of Europe Official Statement. Genetic screenin European proposals. *IME Bull* August 1989:10–11.
9 Houston MJR, Field PA. Practices and policies in th initiation of breastfeeding. *J Obstet Gynecol Neonat Nurs* 1988;**17**:418–24.
10 Faden RR, Chwalow AJ, Orel-Crosby E, *et al*. Wh participants understand about a maternal serum alph fetoprotein screening program. *Am J Public Healt* 1985;**75**:1381–4.
11 Clow CL, Scriver CR. Knowledge about and attitud toward genetic screening among high-school students: th Tay-Sachs experience. *Pediatrics* 1977;**59**:86–91.
12 Cobb S. Social support as a moderator of life stres *Psychosom Med* 1976;**38**:300–14.
13 Field PA. Effectiveness and efficacy of antenatal care. *Mi wifery* 1990;**6**:215–23.

Prenatal screening for cystic fibrosis: psychological effects on carriers and their partners

M E Mennie, M E Compton, A Gilfillan, W A Liston, I Pullen, D A Whyte, D J H Brock

Abstract

This study aimed to assess the psychological impact of screening for cystic fibrosis (CF) carrier status in a population of pregnant women. A cohort of 1798 women, who accepted the offer of testing before 18 weeks of pregnancy, filled in a self administered questionnaire seeking information on their perceived risk of carrier status and their emotional response, as well as a general health questionnaire (GHQ). Sixty-four women identified as CF carriers had partners who received a negative test result. This group and their partners were assessed, together with selected controls, on four further occasions: (1) on receiving the carrier's positive test result; (2) on receiving the partner's negative test result; (3) six weeks later; (4) six weeks after delivery. The instruments used were the GHQ and the Symptom Rating Test (SRT).

When compared to control subjects, carriers showed a significant increase in generalised psychological disturbance which could be attributed specifically to symptoms of anxiety and depression during the period (average four days) that they awaited their partner's test result. On receiving a partner's negative test result, the carriers returned to control levels and maintained this equilibrium. Although there was no significant difference in generalised psychological disturbance between partners and their selected controls, partners did become significantly more anxious and manifested feelings of inadequacy while awaiting their own test result. Both male partners and male control subjects were more likely to become anxious if their partner was distressed.

(*J Med Genet* 1993;30:543–8

Human Genetics Unit, Department of Medicine, University of Edinburgh, Western General Hospital, Edinburgh EH4 2XU, UK.
M E Mennie
M E Compton
A Gilfillan
D J H Brock

Simpson Memorial Maternity Pavilion, Edinburgh, UK.
W A Liston

Department of Psychiatry, Royal Edinburgh Hospital, Edinburgh, UK.
I Pullen

Department of Nursing Studies, University of Edinburgh, Edinburgh, UK.
D A Whyte

Correspondence to Dr Mennie.

Received 2 December 1992. Revised version accepted 10 May 1993.

Population carrier screening for cystic fibrosis (CF) has become possible since the identification of the cystic fibrosis gene[1] and the discovery that a relatively small number of mutations account for the majority of CF chromosomes in the UK population.[2,3] One objective of carrier screening is to provide information to individual persons or couples so that they can make reproductive plans for the future. Another objective is to allow pregnant women to avoid the birth of a child with CF.

There are a number of possible approaches to CF carrier screening and The Cystic Fibrosis Trust has funded three trials to assess the feasibility of delivering screening by alternative routes.[4,5] In Edinburgh a prenatal approach has been adopted.[5] The principal steps in this trial are: (1 to offer pregnant women, attending the antenatal booking clinics of a major maternity hospital, CF carrier screening by way of a mouthwash sample, and to test for six mutations accounting for 85% of CF chromosomes,[3] (2 to offer the partners of women who test positive for the CF gene a carrier test, and (3) to offer prenatal diagnosis to heterozygous couples.

Many of the arguments about CF carrier screening concern the incompleteness of screening. Because the test fails to account for 15% of CF mutations, couples in which neither partner has an identifiable mutation have a residual 1 in 104 000 risk of having an affected child. However, in approximately 4% of couples the women will test positive and her partner negative. These couples face a 1 in 640 risk of having a CF child, substantially greater than their starting risk of 1 in 2500. This study aimed to measure the psychological impact of prenatal CF carrier testing on those couples faced with this intermediate risk.

Subjects and methods

Women up to 18 weeks' gestation presenting for antenatal care at the Simpson Memorial Maternity Pavilion, Edinburgh, were eligible for inclusion in the screening trial. Details of the recruitment and screening method are given elsewhere.[5] The protocol and administration of psychological tests is outlined in fig 1.

From May 1991 to January 1992 a total of 1798 women was screened and 69 (4%) were identified as CF carriers. In all cases the male partner was screened. Excluded from this study were three couples where both partners proved heterozygous. Further exclusions were one couple who suffered a pregnancy loss, one couple who failed to complete the questionnaires, and one screened partner who requested exclusion from the study even though his carrier partner expressed a wish to be included. A total of 64 carrier women and 63 male partners entered the study. One couple subsequently separated resulting in only 62 partners completing the final questionnaires.

For each carrier, two control subjects of the same parity were selected. Control subjects had attended the same antenatal booking clinic

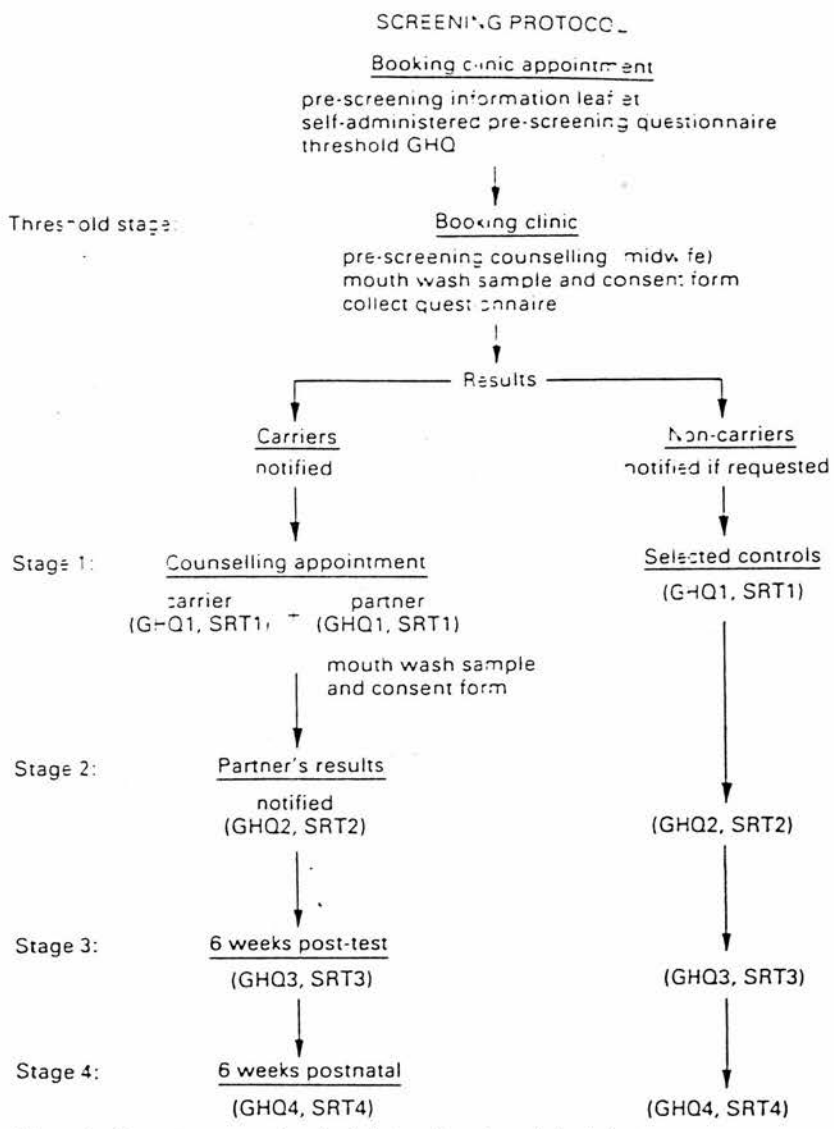


Figure 1 Screening protocol and administration of psychological tests.

as the carrier, had received a negative test result, and had a male partner willing to act as a control subject. A total of 116 female controls and 115 male controls agreed to participate. Of these, 13 couples failed to complete all the questionnaires and two suffered a pregnancy loss. A total of 101 female controls and 100 male controls completed the study to stage 3. Failure to trace two couples resulted in 99 female controls and 98 male controls completing the final questionnaires.

Measures

Sociodemographic data were obtained from subjects' antenatal records. A self administered pre-screening questionnaire assessed perceived carrier risk and emotional response to screening. Threshold psychological assessment and the impact of screening on carriers was assessed by two self rating questionnaires, the General Health Questionnaire (GHQ) and the Symptom Rating Test (SRT).

The GHQ[7] is a screening tool which identifies two main classes of problem: inability to carry out one's normal 'healthy' functions and the appearance of new phenomena of a distressing nature. Given that time is at a premium in an antenatal booking clinic the shortest version, the 12 item GHQ, was chosen using the 3/4 cut off point.[7]

The nature of psychological response was assessed by the SRT.[8] Composed of 30 symptoms briefly defined in simple language, the SRT allows separate scores to be derived for anxiety, depression, inadequacy, and somatic symptoms. Unlike the GHQ, it is not a case finding instrument but will measure psychological distress and is very sensitive to change.

The GHQ scored respondents as positive or negative. The significance of differences between groups was evaluated by the χ^2 test. The SRT ascribed scores to subjects. As the scores were not normally distributed (skew to higher values), the significance of differences between groups was assessed by the median test.

Procedure

An information leaflet was sent to all antenatal patients with their booking clinic appointment. Details of the leaflet have been described previously.[9] Enclosed with the leaflet was a pre-screening questionnaire incorporating a GHQ (termed threshold GHQ). Thus, women suffering from psychological disturbance before receiving a positive CF test result could be ascertained. Women were invited to complete the questionnaire at home and to bring it with them to the clinic. Those women entering the trial who had not completed the questionnaire at home were asked to complete one at the clinic.

Pre-screening counselling and obtaining a mouthwash sample for DNA analysis was carried out by the midwife responsible for booking the patient. Activity at the clinic limited data collection, so only women presenting with a positive GHQ were asked to complete a SRT (termed threshold SRT) to determine the nature of their distress. These women were interviewed by a genetic nurse to ascertain the likely source of their psychological disturbance. GHQ and SRT scores along with interview data were stored on a computer database for ease of storage and recall when a carrier was ascertained.

Women identified as CF carriers were contacted a week later by telephone or, in a minority of cases, by letter and invited to attend the hospital for counselling along with their partner. The couple were seen by a genetic nurse who, before counselling, asked each partner to complete a GHQ and a SRT (termed GHQ1 and SRT1). Counselling was carried out using visual aids and couples were given a detailed information leaflet with a contact telephone number. A consent form and a mouthwash sample for DNA analysis were obtained from the partner.

On receipt of the partner's negative test result (average four days) the couple were contacted in all cases by telephone and informed of the result. A letter was sent confirming the partner's negative test result and reiterating the residual risk of 1 in 640 of having an affected child. Enclosed were a stamped addressed envelope and a GHQ and a

Table 1 Sociodemographic data (percentages in brackets).

	Total population n = 1798	Carriers n = 64	Selected controls (n = 101
Age (y)			
Mean	28·07	27·86	28·64
Range	16–44	18–44	20–40
Parity			
Primiparous	916 (51	35 (55	52 (51
Multiparous	882 (49	29 (45	49 (49
Gestation wk			
Mean	12·25	11·94	12·25
Range	6–18	7–16	7–15
Marital status			
Married	1316 (73	48 (74	82 (81
Single	409 (23	14 (22	16 (18
Divorced	46 (3	0 (0	3 (3
Separated	24 (1	1 (2	0 (0
Widowed	3 (–	1 (2	0 (0
Social class			
1	227 (13	5 (8	11 (11
2	553 (31	15 (25	31 (31
3	601 (33	23 (36	36 (35
4	158 (9	10 (16	16 (16
5	91 (5	4 (6	3 (3
Unemployed	141 (8	4 (6	3 (3
Student	27 (1	0 (0	1 (1

SRT (termed GHQ2 and SRT2). Six weeks later the couple were sent a further postal GHQ and a SRT (termed GHQ3 and SRT3) and finally six weeks after delivery the same two measures were sent (termed GHQ4 and SRT4).

Control couples received a postal GHQ and a SRT at comparable intervals to carriers and partners.

Table 2 Perception of carrier risk and emotional response to screening (percentages in brackets).

Perception of risk	Emotional response*			
	Anxious	Reassured	Don't know	Total
1 in 4	78 (24)	154 (47)	97 (29)	329 (100)
1 in 25	258 (24)	533 (51)	264 (25)	1055 (100)
1 in 100	7 (35)	13 (65)	0	20 (100)
1 in 200	1 (6)	11 (69)	4 (25)	16 (100)
Don't know	78 (21)	169 (45)	129 (34)	376 (100)

* Two subjects no response.

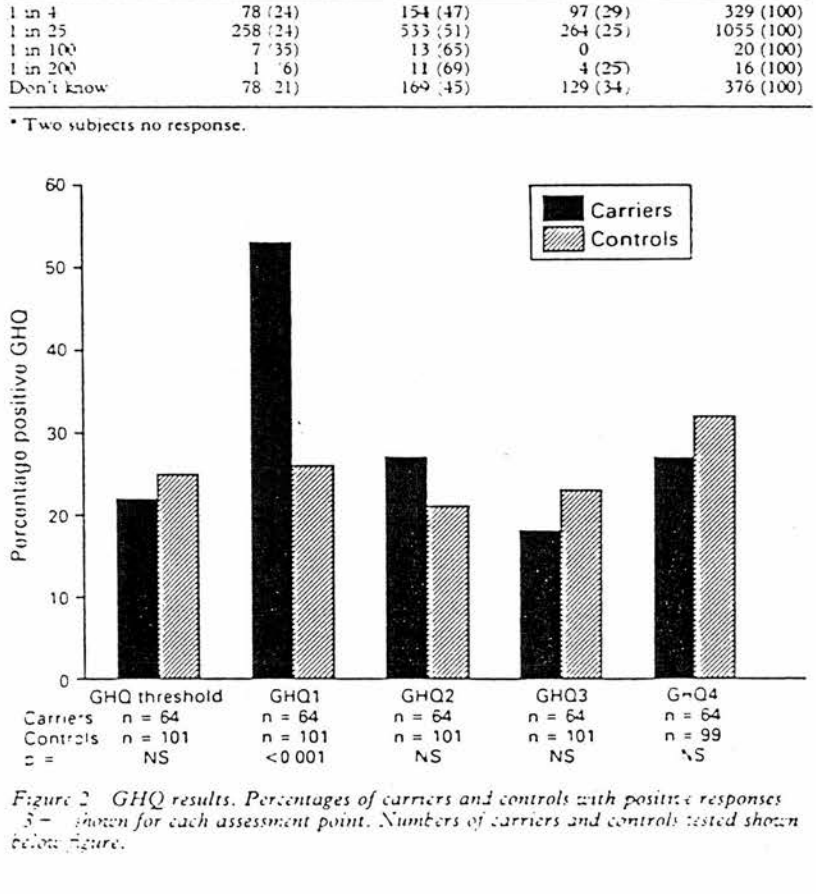


Figure 2 GHQ results. Percentages of carriers and controls with positive responses (5–) shown for each assessment point. Numbers of carriers and controls tested shown below figure.

Results

SOCIODEMOGRAPHIC DATA

The sociodemographic characteristics of th screened population, carriers, and controls ar shown in table 1. The sample was weighte toward the higher socioeconomic group (assessed from the occupation of the head o the household using the Registrar General classification). This reflects the tendency fo women of the higher social classes to presen earlier for antenatal care and to have a stab partnership. Furthermore, a majority o women from a sizeable area housing those o the lower socioeconomic groups booked fo antenatal care at a clinic within that area.

ANXIETY AND PERCEIVED CARRIER RISK

The pre-screening questionnaire assessed pe ceived carrier risk. Five carrier risk option were stated and women were asked to sele one (table 2). Of the 1798 women screened 1055 (59%) perceived their risk correctly. substantial number (378, 21%) had no percep tion of their risk, and a minority (36, 2% perceived their risk to be considerably lowe than the 1 in 25 risk stated in the pre-screenin information leaflet. A further 329 (18%) pe ceived their risk to be 1 in 4.

Asked to indicate their emotional respons to being screened, 880 (49%) stated they we reassured and 494 (28%) did not know ho they felt. Anxiety was felt by 422 (23%) women and there was no response in two case There was no correlation between anxie about being screened and perceiving one's ris to be higher than 1 in 25 (table 2).

RESULTS ON THE GENERAL HEALTH QUESTIONNAIRE (GHQ)

Preliminary results of GHQ data on carri and control subjects have been reported els where.[5] In this extended study a total of 57 (32%) of the screened population presente with a positive threshold GHQ. Many wome (44%) cited symptoms of pregnancy as th problem. Twelve percent stated their preg nancy was unplanned, 10% had a poor obst tric history, and a further 7% felt general anxious about the pregnancy. Two perce were worried about other antenatal diagnost tests but only two women were concerne about the CF screening test. Four percent women had a psychiatric history.

Of the 64 carriers, 14 (22%) had a positi GHQ against 25 (25%) among the contro (fig 2). On receiving positive test results, t proportion of carriers (53%) with a positi GHQ1 score was significantly greater th the proportion of control subjects (27%) ($p < 0{\cdot}001$). At the time of their partner's neg tive test result (GHQ2), at six weeks after t test (GHQ3) and at six weeks after delive (GHQ4), carriers showed no significant di ference in the proportion of positive scor when compared to selected controls (fig 2).

No significant difference was found betwe the proportion of partners and their select controls with a positive GHQ score at any

the four assessment points (fig 3). However, 14 of 15 (93%) partners with a positive GHQ1 had a female partner who had also scored positive. Nine of the 10 (90%) male controls who had positive GHQ1 scores had a female partner with a positive GHQ score. Males were, therefore, significantly more likely to present with psychological distress if their female counterpart was also distressed (χ^2, $p < 0.001$).

SYMPTOM RATING TEST (SRT)

Five hundred and nineteen of the 576 (90%) of the total screened population with a positive threshold GHQ were interviewed and completed a threshold SRT. Among this group were 14 (22%) carriers and 25 (25%) selected controls. There was no significant difference in the threshold SRT scores of the three groups. However, at SRT1 (carrier receiving positive test result) there was a significant difference between carriers and controls in the total score for generalised psychological disturbance (median test, $p < 0.005$) and specifically in the subscores for anxiety and depression (median test, $p < 0.001$, table 3).

On receiving their partner's negative test result (SRT2) the scores of carriers returned to control levels and remained there at the six week post-test point (SRT3) and again at the six week post-delivery point (SRT4) (table 3).

There was no significant difference between SRT scores for generalised psychological disturbance of the partners of carriers when compared with their selected controls (table 4). Anxiety and inadequacy subscores were significantly higher than those of controls at the time when carriers were given their positive test results (median test, $p < 0.05$ and $p < 0.02$ respectively, table 4).

There was a significant decrease in the subscores of anxiety in SRT1 and SRT2 scores in both carriers and their partners (table 5). This also showed up in the subscores for depression among carriers.

Table 3 Median of symptom rating test (SRT) scores of carriers and controls.

SRT1	Carriers (n = 46)	Controls (n = 101)	
Total	11·5	7·0	p < 0·005
Anxiety	4·5	1·0	p < 0·001
Depression	4·0	2·0	p < 0·001
Somatic	1·0	1·0	NS
Inadequacy	2·5	2·0	NS
SRT2	Carriers (n = 64)	Controls (n = 101)	
Total	7·0	7·0	NS
Anxiety	2·0	1·0	NS
Depression	2·0	2·0	NS
Somatic	1·0	1·0	NS
Inadequacy	2·0	2·0	NS
SRT3	Carriers (n = 64)	Controls (n = 101)	
Total	5·0	7·0	NS
Anxiety	1·0	1·0	NS
Depression	2·0	2·0	NS
Somatic	1·0	2·0	NS
Inadequacy	2·0	2·0	NS
SRT4	Carriers (n = 64)	Controls (n = 99)	
Total	6·0	7·0	NS
Anxiety	1·0	1·0	NS
Depression	2·0	2·0	NS
Somatic	0·0	0·0	NS
Inadequacy	2·0	2·0	NS

Table 4 Median of symptom rating test (SRT) scores of partners and controls.

SRT1	Partners (n = 63)	Controls (n = 100)	
Total	5·0	3·0	NS
Anxiety	2·0	1·0	p < 0·05
Depression	1·0	1·0	NS
Somatic	0·0	0·0	NS
Inadequacy	1·0	0·0	p < 0·02
SRT2	Partners (n = 63)	Controls (n = 100)	
Total	3·0	2·0	NS
Anxiety	1·0	0·0	NS
Depression	1·0	1·0	NS
Somatic	0·0	0·0	NS
Inadequacy	1·0	0·0	NS
SRT3	Partners (n = 63)	Controls (n = 100)	
Total	2·0	2·5	NS
Anxiety	0·0	0·5	NS
Depression	0·0	1·0	NS
Somatic	0·0	0·0	NS
Inadequacy	1·0	0·0	NS
SRT4	Partners (n = 62)	Controls (n = 98)	
Total	3·0	3·0	NS
Anxiety	0·0	0·0	NS
Depression	1·0	1·0	NS
Somatic	0·0	0·0	NS
Inadequacy	1·0	1·0	NS

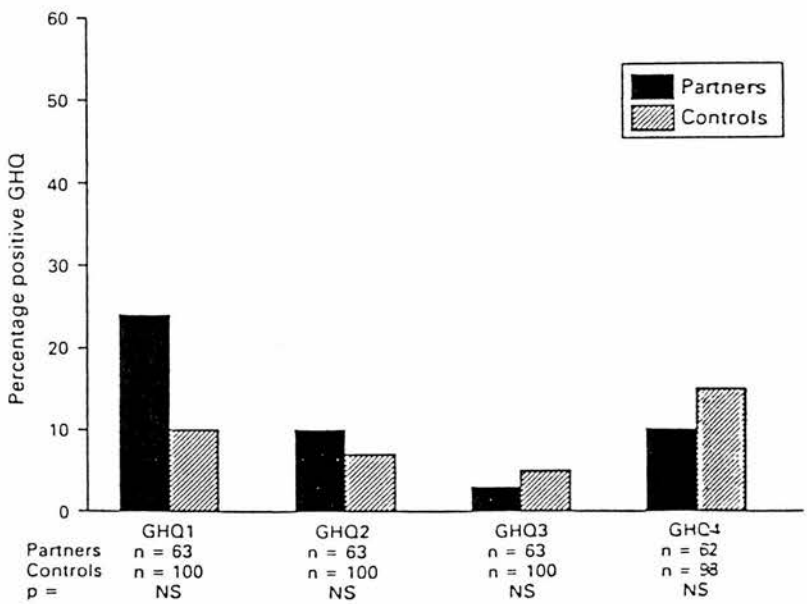


Figure 3 GHQ results. Percentages of partners and controls with positive responses (3−) shown for each assessment point. Numbers of partners and controls tested shown below figure.

Discussion

The findings from this study show that pre natal carrier screening delivered in two stage does generate some psychological disturbance In the identified carrier there is a significan

Table 5 Comparison of median of SRT1 and SRT2 scores in carriers (n = 64) and partners (n = 63).

Carrier's scores	SRT1	SRT2	
Total	11·5	7·0	NS
Anxiety	4·5	2·0	p < 0·001
Depression	4·0	2·0	p < 0·02
Somatic	1·0	1·0	NS
Inadequacy	2·5	2·0	NS
Partner's scores	SRT1	SRT2	
Total	5·0	3·0	NS
Anxiety	2·0	1·0	p < 0·005
Depression	1·0	1·0	NS
Somatic	0·0	0·0	NS
Inadequacy	1·0	1·0	NS

increase in generalised psychological disturbance, specifically anxiety and depression, when compared to a control population. This reaction occurs in response to learning of their carrier status and lasts for the period (approximately four days in this study) awaiting their partner's test result. On receiving their partner's negative test result the distress subsides and there is no indication of a resurgence during the pregnancy or in the immediate postnatal period. Although the longer term effects are unknown, three carriers from this study have subsequently embarked upon a further pregnancy.

Male partners manifested symptoms of anxiety and inadequacy during the period awaiting their test result, but this disappeared on receipt of a negative result. Both partners and male control subjects were significantly more likely to manifest psychological disturbance if their female partner was distressed.

Thirty-two percent of women presented at the antenatal booking clinic with a positive GHQ score. This is comparable with other studies.[10] Previous work has shown that randomly selected samples from the community will contain quite high proportions of persons with degrees of psychological disturbance ranging from mild to severe.[11] Indeed it is estimated that 25% of patients seen in general practice have anxiety as a clinically significant component of their condition.[12]

In this study subjects presenting with negative GHQ scores submitted mean SRT scores comparable with normal subjects in previous studies.[8 13] Carriers with positive GHQ1 scores submitted SRT1 scores well below those reported for psychiatric patients[8 14] and comparable with scores generated by the Symptom Questionnaire in patients undergoing amniocentesis.[14] This is a point worth noting as previous studies have suggested that levels of anxiety in pregnant women who receive a positive test result can be extremely high, above those for psychiatric patients.[15] It is clear that a substantial number of women will enter a prenatal screening programme with concurrent stress. Indeed, five of the 14 carriers who presented with a positive threshold GHQ score maintained these scores throughout; for reasons of recent bereavement (three cases), diagnosis of chronic illness in the partner (one case), and regular ECG monitoring throughout pregnancy for attacks of breathlessness (one case). Eleven out of 25 control subjects who presented with positive threshold scores maintained these scores throughout the study for a variety of reasons. Threshold psychological assessment on all women screened served not only to ensure for the purposes of this study that there was no significant psychological difference between carriers and control subjects at the outset, but proved valuable in the wider screening trial for ascertaining women identified as CF carriers who were already experiencing stress and might require extra counselling and support.

Previous studies on patients undergoing prenatal screening have indicated that once a woman perceives her pregnancy has been threatened she continues to be concerned.[16] Conversely, others have shown a dramatic return to normal once a negative test result is given.[14 17 18] The results of this study agree with the findings of the latter. A notable effect of prenatal CF carrier screening upon two carriers was their subsequently declining α fetoprotein (AFP) screening. A further two carriers received abnormal AFP screening results and underwent amniocentesis for chromosome studies. Despite this only one of these four women was included in the 6% of carriers who stated they were against the test being offered routinely to pregnant women. A concern must be that some women may not be so resilient to multiple provoking agents during pregnancy, which could foreseeably occur if a woman experiences several positive screening test results, and particularly were she already suffering from concurrent stress. A positive aspect is that the developing area of prenatal screening will help us to consider more carefully the whole area of psychosocial support in the provision of antenatal care, which some perceive to have been neglected.[19]

Psychological disturbance is considered a normal rather than a pathological response to prenatal diagnosis.[20] It has been suggested that counselling efforts should support the person's attempt to cope with stress accompanying the procedure rather than not provide screening.[21] The results of this study suggests that stress resulting from being identified as a CF carrier during pregnancy is short lived and that those couples faced with an intermediate risk of having a CF child cope satisfactorily.

We thank the antenatal patients who responded to this study and the medical and midwifery staff of the Simpson Memorial Maternity Pavilion for making this trial possible. The study was funded by grants from The Cystic Fibrosis Trust and the Scottish Home and Health Department.

1 Kerem BS, Rommens JM, Buchanan JA, *et al.* Identification of the cystic fibrosis gene: genetic analysis. *Science* 1989;245:1073–80.
2 Watson EK, Mayall ES, Simova L, *et al.* The incidence of delta F508 mutation, and associated haplotypes, in a sample of English CF families. *Hum Genet* 1990;85:435–6.
3 Shrimpton AE, McIntosh I, Brock DJH. The incidence of different cystic fibrosis mutations in the Scottish population: effects on prenatal diagnosis and genetic counselling. *J Med Genet* 1991;28:317–21.
4 Watson EK, Mayell E, Chapple J, *et al.* Screening for carriers of cystic fibrosis through primary health care services. *BMJ* 1991;303:504–7.
5 Mennie ME, Gilfillan A, Compton M, *et al.* Prenatal screening for cystic fibrosis. *Lancet* 1992;340:214–16.
6 Goldberg D, Williams P. *A user's guide to the General Health Questionnaire.* Windsor: NFER-Nelson, 1988.
7 Mari J, Williams P. A comparison of the validity of two psychiatric screening questionnaires GHQ12 and SRQ20 in Brazil, using relative operating characteristic analysis. *Psychol Med* 1985;15:651–9.
8 Kellner R, Sheffield BF. A self-rating scale of distress. *Psychol Med* 1973;3:88–100.
9 Mennie M, Liston WA, Brock DJH. Prenatal cystic fibrosis carrier testing: designing an information leaflet to meet the specific needs of the target population. *J Med Genet* 1992;29:308–12.
10 Sharp D. Validation of the 30-item General Health Questionnaire in early pregnancy. *Psychol Med* 1988;18:505–7.
11 Goldberg D, Huxley P. *Common mental disorders. A bio-social model.* Tavistock: Routledge, 1992.
12 Wilkinson G. Anxiety: recognition and treatment in general practice. Oxford: Radcliffe Medical Press Ltd, 1992.
13 Cochrane R. A comparative evaluation of the Symptom Rating Test and the Langer 22-item Index for use in epidemiological surveys. *Psychol Med* 1980;10:115–24.
14 Fava G, Kellner R, Perini G, *et al.* Italian validation of the

Symptom Rating Test (SRT) and Symptom Questionnaire. *Can J Psychiatry* 1983;28:117–23.
15 Robinson JO, Hibbard BM, Laurence KM. Anxiety during a crisis: emotional effects of screening for neural tube defects. *J Psychosom Res* 1984;28:163–9.
16 Tabor A, Jonsson MH. Psychological impact of amniocentesis in low risk women. *Prenat Diagn* 1987;7:443–9.
17 Burton BK, Dillard RG, Clark EN. The psychological impact of false positive elevations of maternal serum alpha-fetoprotein. *Am J Obstet Gynecol* 1985;151:77–82.
18 Tsoi MM, Hunter M, Pearce M. Ultrasound scanning in women with raised serum alpha-fetoprotein: short term psychological effects. *J Psychosom Res* 1981;31:35–9.
19 Campbell IE, Field PA. Common psychological concerns experienced by parents during pregnancy. *Can Ment Health* 1988;37:2–5.
20 Blumberg B. The emotional implications of prenatal diagnosis. In: Emery AEH, Pullen I, eds. *Psychological aspects of genetic counselling*. London: Academic Press, 1988: 213–7.
21 Childs B, Greidis L, Kaback M, Kazazian HH, Tay-Sachs screening: social and psychological impact. *Am J Hum Genet* 1976;28:550–4.

Clin Genet 1993: 44: 102–106
Printed in Denmark . All rights reserved

CLINICAL GENETICS
ISSN 0009-9163

Genetics and Society

Prenatal screening for cystic fibrosis: attitudes and responses of participants

Mennie M, Compton M, Gilfillan A, Axton RA, Liston WA, Pullen I, Whyte D, Brock DJH. Prenatal screening for cystic fibrosis: attitudes and responses of participants.
Clin Genet 1993: 44: 102–106. © Munksgaard, 1993

A screening programme to detect cystic fibrosis heterozygotes has been running in the antenatal clinics of a major Edinburgh maternity hospital for more than 2 years. A questionnaire was used to assess participants' knowledge of the genetics of the disorder and their attitudes to being screened. The respondents were 64 female heterozygotes and 63 of their non-heterozygous male partners, 101 female controls and 100 male controls. Although the two groups of controls received far less direct information than the carriers and their partners, all four groups were well informed about the genetics of cystic fibrosis and the significance of being a gene carrier. A majority of each group felt that adequate information had been given in the information leaflet, that they understood the purpose of screening and that they were glad to have participated. There was a consensus that CF carrier testing should be routinely offered to pregnant women, and also that it should be available in family planning clinics and GP health centres, but not in schools.

Moira Mennie, Mary Compton, Annette Gilfillan, R. A. Axton, W. A. Liston, I. Pullen, Dorothy Whyte and D. J. H. Brock

Human Genetics Unit, University of Edinburgh, Western General Hospital, Edinburgh, UK

Key words: antenatal clinic– attitudes to screening – cystic fibrosis – heterozygotes – partners – pregnancy

Professor D. J. H. Brock, University Department of Human Genetics, Western General Hospital, Edinburgh EH4 2XU, UK

Received 23 February, revised version received and accepted for publication 5 April 1993

The cloning of the cystic fibrosis (CF) gene (Rommens et al. 1989), together with the demonstration that up to 85% of mutant alleles may be detected relatively easily (Shrimpton et al. 1991), has made it possible to institute programmes of population heterozygote screening. Amongst various models for delivery of such programmes, those using antenatal clinics have obvious attractions (Mennie et al. 1992a, b). Pregnant women and their partners are highly motivated to establish their heterozygote status and, through prenatal diagnosis, to avoid the risk of delivering a CF child. Although testing during pregnancy generates some stress, it is comparatively short-lived and effectively disappears once partners have been shown to be negative for detectable CF alleles (Mennie et al. 1992b, 1993).

The absence of significant anxiety amongst the participating population, though important, does not necessarily make a screening programme valid. It is essential to have some idea of the attitude of mind of carriers and their partners, their understanding of the processes involved and their response to the discovery of their genetic status. In this study we have assessed these factors by a specifically designed, self-administered questionnaire.

Materials and methods

Subjects

Women booking for delivery of their babies at the Simpson Memorial Maternity Pavilion, Edinburgh, were invited to enter a two-step prenatal screening trial for CF. Details of the protocol and recruitment methods for this trial have been published previously (Mennie et al. 1992a, b).

Between May 1991 and January 1992, 69 women were identified as CF carriers amongst a total of 1798 screened. In each case the male partner was also screened, and amongst the 69 there were three 1 in 4 risk couples. These three couples were excluded from this study, along with one couple whose pregnancy failed to continue, and a further couple and one male partner who failed to complete the questionnaire. A total of 64 carriers and 63 partners participated in the study.

Two control subjects were selected for each carrier. The controls had attended the same booking clinic as the carrier, were of the same parity and had received a negative test result. The male partners of these female controls were also invited to participate in the study, and served as controls for the partners

carriers. In all, 116 female controls and 115 male ntrols agreed to take part, of whom 13 couples did)t respond to the questionnaire, while a further two uples had pregnancies which failed to continue. A tal of 101 female controls and 100 male controls mpleted the questionnaires.

ethods

ogether with their booking clinic appointment, l antenatal patients attending the Simpson Me- iorial Maternity Pavilion received a leaflet outlin- ig the aims of prenatal CF carrier testing and escribing the procedure. The designing of the leaf- :t has been described (Mennie et al. 1992a). The :aflet stated the CF carrier frequency, displayed ie mode of inheritance of CF in diagrammatic orm, and emphasized that carrier couples had a 1 4 risk of an affected child. It pointed out that a amily history of CF was not a prerequisite to eing a CF carrier. Care was taken to stress the act that being a carrier was unimportant unless ie partner was also a carrier.

Women identified as CF carriers were invited to ttend the hospital with their partner for coun- elling. An additional leaflet was given to all carriers and their partners reiterating in greater detail the pre-screening information. This leaflet gave the risk of heterozygosity amongst the siblings of carriers, recommended that relatives should be screened before pregnancy and advised how to go about this. Partners received test results after an average of 4 days' wait. After 6 weeks, carriers and their negative partners were sent a "facts and feelings" questionnaire by post, with a stamped addressed envelope for return. The "facts" section was composed of six statements derived from the pre-screening information booklet. Subjects were asked to tick which statements they thought to be true. The "feelings" section of the questionnaire was composed of a further six statements designed to assess subject's attitudes towards the prenatal screening trial specifically, and also towards CF carrier testing in general.

Sociodemographic data were obtained from the hospital antenatal records. Significance was assessed by the χ^2 test with Yates' correction.

Results

Sociodemographic data showed no significant differences between carriers and controls or between

ble 1. Percentage of true and false responses to "facts" questionnaire (part . No one failed to respond

estion	True	False	
Any couple can have a child with CF			
female carriers	39	61	
female controls	41	59	
male partners	43	57	
male controls	42	58	
A couple can have a child with CF if:			
a) Only one partner carries a single CF gene			
female carriers	11	89	
female controls	10	90	
male partners	13	87	
male controls	18	82	
b) If both partners carry a single CF gene			
female carriers	95	5	
female controls	92	8	
male partners	98	2	*
male controls	84	16	*
One in 25 people in Britain carry a single CF gene			
female carriers	100	0	+
female controls	77	23	+
male partners	92	8	*
male controls	74	26	*
Even if you have no family history of CF you can carry a single CF gene			
female carriers	91	9	
female controls	94	6	
male partners	89	11	
male controls	96	4	

test on case versus control; * $p < 0.01$, + $p < 0.001$.

Table 2. Percentage responses to "facts" questionnaire (part 2). No one failed to respond

Question

5. If both partners carry a single CF gene their chance of having a child with CF is:
   a) 1 in 2
   b) 1 in 4
   c) 1 in 20
   d) All their children will have CF

Response

	a	b	c	d	
Female carriers	2	98	0	0	
Female controls	0	94	6	0	
Male partners	2	97	1	0	*
Male controls	4	81	14	1	*

Question

6. If you carry a single CF gene this means:
   a) Your health will be affected
   b) You will develop the disease CF
   c) It is only important if your partner carries a single CF gene

Response

	a		b		c		
	True	False	True	False	True	False	
Female carriers	0	100	0	100	100	0	
Female controls	0	100	1	99	96	4	
Male partners	0	100	0	100	100	0	
Male controls	1	99	2	98	93	7	

χ^2 test on cases versus controls; * $p < 0.01$.

Table 3. Percentages responding to "feelings" questionnaire (part 1)

Question 1: I feel that the information I/my partner was given about the CF carrier test (a) before the antenatal clinic (b) at the antenatal clinic was about right, too much or not enough

(a) Before the antenatal clinic

	About right	Too much	Not enough
Female carriers	62	2	35
Female controls	65	0	35
Male partners	62	2	36
Male controls	54	0	46

(b) At the antenatal clinic

	About right	Too much	Not enough
Female carriers	75	2	23 } *
Female controls (1 no response)	88	0	12 } *
Male partners	87	2	11
Male controls (1 no response)	87	2	11

Question 2: I feel that I understood what the CF test was all about before I/my partner was tested.

	Yes	No/don't know
Female carriers	70	30
Female controls	83	17
Male partners	54	46
Male controls	60	40

Question 3: I am glad that I/my partner had the CF carrier test

	Yes	No/don't know
Female carriers	80	20 } *
Female controls	97	3 } *
Male partners	90	10
Male controls	98	2

Question 4: I feel that the CF carrier test should routinely be offered to pregnant women

	Yes	No/don't know
Female carriers	88	12
Female controls	96	4
Male partners	94	6
Male controls	94	6

χ^2 test on cases versus controls; * $p < 0.001$.

male partners and their respective controls in any of the factors examined.

The six questions in the "facts" questionnaire are shown in Tables 1 and 2, along with the responses. The most significant differences between cases (i.e. carriers or their partners) and their respective controls are to be seen in question 3, the population frequency of CF heterozygosity, with almost a quarter of controls of both sexes misidentifying the correct figure. Controls of male partners were also significantly different to their cases in response to questions 2b and 5. It must also be noted that the unexpected range of response to question 1 suggests imprecision.

Table 4. Percentages responding yes to "feelings" questionnaire (part 2). No one failed to respond

Question 5: I feel that I am in favour of testing for CF carriers in:

	Schools	Family planning clinics	GP health centres
Female carriers	30	86	89
Female control	36	89	93
Male partners	32	81	87
Male controls	38	81	89

Question 6: If I turned out to carry a CF gene, I feel I would tell:

	My partner	My sibs	My children	Other relatives	Friends
Female carriers	100	92 } *	92 } *	59	45
Female controls	100	78 } *	78 } *	53	31
Male partners	100	79 } *	87	49	30
Male controls	100	63 } *	79	34	22

χ^2 test on cases versus contorls (agreement/non-agreement); * $p < 0.05$.

Questions and responses to the "feelings" questionnaire are shown in Tables 3 and 4. Carriers were significantly more likely than controls to feel that the information given at the antenatal clinic was insufficient (question 1b), and to have ambivalent feelings about having been screened (question 3). A high proportion of each group (88–96%) felt that the CF carrier test should routinely be offered to pregnant women (question 4). There was also strong support for carrier screening in family planning clinics and GP health centres, but not in schools (question 5). Carriers and their partners were significantly more likely than their respective controls to tell their sibs and their children that they carried a CF gene (question 6).

Discussion

In this study we have attempted to assess two important aspects of a screening programme: the understanding by the participants of the genetic facts of the disease and their feelings about having agreed to take part. The four groups involved had rather different access to information and also different experiences of the programme. Those identified as carrying mutant CF alleles and their partners had experienced some anxiety while awaiting the partner's test result (Mennie et al. 1992b, 1993). Both carriers and partners had been counselled in one-on-one sessions with a genetic nurse and had received additional written information. In contrast, the two control groups would have derived most of their knowledge of CF from the infor-

mation leaflet sent out at the time of their booking appointment, 2 to 3 months before they completed the "facts" and "feelings" questionnaire. Although the information leaflet was presumably still available for consultation, it is gratifying that in some sections of the "facts" questionnaire all four groups had near maximum scores. This is seen in the response to whether it is possible to carry a CF gene if you have no family history of CF (question 4, Table 1) and in the response to the question about the effect of CF heterozygosity on general health (question 6, Table 2). Concern has been raised that being a carrier of a recessive gene may cause an individual to have a less positive view of his or her health (Marteau et al. 1992).

Three of the four groups had an excellent idea of the risk of having a child with CF when both partners carried a mutant gene (question 5, Table 2). However, even though controls of the female carriers were well informed on this point, the male controls of their partners proved fallible. An earlier study found that only 63% of males had read the prescreening leaflet and had participated in the screening decision (Mennie et al. 1992a).

We were not surprised that controls scored poorly in comparison to cases in the question about CF heterozygosity frequency in Britain (question 3). They had far less access to this information, and as they screened negative, it no longer really concerned them. Similar findings have been reported by others (Watson et al. 1992).

Responses to the "facts" questionnaire also allowed us to focus on shortcomings in our information leaflets. Approximately 40% of all groups thought the statement "any couple can have a child with CF" to be true (question 1, Table 1). Although the leaflet showed in diagrammatic form that each partner had to be a carrier, it also emphasized that current heterozygote tests are incomplete. Thus in a literal sense the statement is true, even though we expected respondents to find it false. This illustrates how easy it is to introduce ambiguities into a questionnaire. Field & Renfrew (1991) have criticized the failure of health care professionals to inspect in detail the content of printed information issued to patients.

From the "feelings" questionnaire, it would seem that there is a desire amongst all four groups for more information on the carrier test before attending the antenatal clinic (question 1a, Table 3). Furthermore, there was a significant difference between carriers and their controls in feelings about whether enough information had been supplied at the antenatal clinic (question 1b, Table 3). A much larger proportion of men than women did not feel that they knew what the CF test was all about before either they or their partners were tested (question 2). Again this may reflect the fact that around 37% of men do not read the prescreening information leaflet nor enter into the decision to participate in the screening programme (Mennie et al. 1992a). Nonetheless, a very large majority of all four groups were glad that they or their partner had taken the CF carrier test (question 3). The only ambivalence appeared amongst the carriers themselves.

There seems to be considerable tolerance for the timing of the CF carrier test. High proportions of all four groups felt that it should be routinely offered in pregnancy, as well as being available at family planning clinics and GP health centres. There was minority support for screening in schools (question 5); yet other studies have found this to be the popular option (Green 1992, Zeesman et al. 1984).

All respondents stated that they would tell their partner if they turned out to be a CF carrier (question 6, Table 4). Carriers and their partners were more likely to tell their sibs, children, other relatives and friends about being a carrier than were their respective controls. This undoubtedly reflects the one-on-one counselling sessions where a family tree was drawn and risks to siblings explained, in addition to giving carriers and their partners a leaflet outlining the screening procedure for relatives.

Loader et al. (1991), studying prenatal screening for haemoglobinopathies, have pointed out that learning during genetic counselling sessions is often below expectations. Problems may also arise when the provider rather than the counsellor initiates the testing, a feature of population screening. It is suggested that videotape presentations and home visits might be advantageous. However, in this programme we present evidence that a simple, carefully-designed leaflet (Mennie et al. 1992a) can be very effective in communicating essential genetic facts. Although all our female controls were given a great deal of information on a variety of topics during the booking-in procedure, in both verbal and written form, they had retained and understood most of the key features of the CF leaflet sent to them several months before they answered the questionnaire. We presume that many re-read the leaflet before attempting to answer questions. Our findings document a high level of understanding and a positive view of the screening procedure.

Acknowledgements

We are grateful to all the patients and their partners who took time to respond to the questionnaire. We thank the medical and nursing staff of the Simpson Memorial Maternity Pavilion for making this trial possible. The study was funded by the

Cystic Fibrosis Trust, the Medical Research Council and the Scottish Office Home and Health Department.

References

Field PA, Renfrew M. Teaching and support: nursing input in the postpartum period. Int J Nurs Stud 1991: 28: 131–144.

Green JM. Principles and practicalities of carrier screening: attitudes of recent parents. J Med Genet 1992: 29: 313–319.

Loader S, Sutera CJ, Walden M, Kozyra A, Rowley PT. Prenatal screening for haemoglobinopathies. II. Evaluation of counselling. Am J Hum Genet 1991: 48: 447–451.

Marteau TM, Van Duijn M, Ellis I. Effects of genetic screening on perceptions of health: a pilot study. J Med Genet 1992: 29: 24–26.

Mennie ME, Liston WA, Brock DJH. Prenatal cystic fibrosis carrier testing: designing an information leaflet to meet the specific needs of the target population. J Med Genet 1992a: 29: 308–312.

Mennie ME, Gilfillan A, Compton ME, Curtis L, Liston WA, Pullen I, Whyte DA, Brock DJH. Prenatal screening for cystic fibrosis. Lancet 1992b: 340: 214–216.

Mennie ME, Compton ME, Gilfillan A, Liston WA, Pullen I, Whyte DA, Brock DJH. Prenatal screening for cystic fibrosis: the psychological effects on carriers and their partners. J Med Genet, in press.

Rommens JM, Iannuzzi MC, Kerem B, Drumm ML, Melmer G, Dean M, Rozmahel R, Cole JL, Kennedy D, Hidaka N, Zsiga M, Buchwald M, Riordan JR, Tsui L-C, Collins FS. Identification of the cystic fibrosis gene: chromosome walking and jumping. Science 1989: 245: 1059–1065.

Shrimpton AE, McIntosh I, Brock DJH. The incidence of different cystic fibrosis mutations in the Scottish population: effects on prenatal diagnosis and genetic counselling. J Med Genet 1991: 328: 317–321.

Watson EK, Mayall ES, Lamb J, Chapple J, Williamson R. Psychological and social consequences of a community carrier screening programme for cystic fibrosis. Lancet 1992: 340: 217–220.

Zeesman S, Clow C, Cartier L, Scriver CR. A private view of heterozygosity: eight-year follow-up study on carriers of the Tay-Sachs gene detected by high school screening in Montreal. Am J Med Genet 1984: 18: 769–778.

05 DEC. 1994

HUMAN GENETICS UNIT
Department of Medicine (WGH)

The University of Edinburgh
Western General Hospital
Edinburgh EH4 2XU

Fax 031 343 1863
Telephone 031 332 7917
031 332 2525 extension 4499

29th November 1994

The Publishers
Clinical Genetics
Munksgaard
International Publishers Limited
35 Norre Sogade
PO Box 2148
DK - 1016
Copenhagan K
Denmark

Dear Sirs

re: Mennie M, Compton M, Gilfillan A, Axton RA, Liston WA, Pullen I, Whyte D, Brock DJH. Prenatal screening for cystic fibrosis: atitudes and responses of participants. Clinical genetics 1993; 44:102-106

I write to ask if you would give me permission to include a photocopy of the above publication in my PhD thesis.

Yours faithfully,

Moira E Mennie

Please excuse the informality. To speed our reply we have answered on your own letter
Munksgaard, 35 Norre Sogade, P O.Box 2148,
DK 1016 Copenhagen K, Denmark.
Telephone +45 33 12 70 30. Fax +45 33 12 93 87

Permission granted by the copyright owner, ~~contingent upon the consent of the author,~~ provided complete credit is given to the original source and © date Munksgaard International Publishers Ltd., Copenhagen, Denmark.

per: – 5 DEC 1994

Vibeke Rosendal Haines
Editorial Assistant
Journals Div.